

Quality and coverage of the NSW  
Register of Congenital Conditions  
using admitted patient data:  
A record linkage study



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# ACRONYMS AND ABBREVIATIONS

APD	NSW public and private admitted patient data
BPA	British Paediatric Association
CHeReL	Centre for Health Record Linkage
CI	Confidence interval
ICD-10-AM	International Statistical Classification of Diseases and Related Health Problems, 10th Revision, Australian Modification
LHD	Local Health District
MLK	Master Linkage Key
NSW	New South Wales
PPN	Project Person Number
RoCC	NSW Register of Congenital Conditions

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# EXECUTIVE SUMMARY

The purpose of this project was to assess the quality and coverage of reporting to the NSW Register of Congenital Conditions (RoCC) compared to admitted patient records. Records of cases reported to the RoCC in 2009 were linked to records of the NSW Admitted Patient Data Collection (APD) indicating a congenital condition eligible to be reported in the same period, and medical records of APD cases that were not linked to the RoCC were reviewed. The enumeration of cases of congenital conditions reported to the RoCC was evaluated and the quality of information on cases reported on the APD was assessed.

This study found that about 30% of cases of congenital conditions were reported to the APD and not to the RoCC. There were 2,187 cases of congenital conditions reported to the RoCC, and the medical record review of APD records confirmed 866 additional cases, giving a total of 3,053 cases for 2009. The additional cases comprised 753 livebirths (87.0%), 109 terminations of pregnancy (12.6%), and 4 stillbirths (0.5%). Of particular interest were the findings for neural tube defects, where 77 cases were reported to the RoCC and the medical record review revealed an additional 43 cases, giving a total of 120 cases for 2009.

The overall sensitivity of the RoCC was 71.6%. The RoCC was highly sensitive (>90%) for chromosomal abnormalities, eye conditions, gastrointestinal conditions and respiratory conditions among liveborn babies. Among terminations of pregnancy, stillbirths and cases with unknown outcome the RoCC was highly sensitive (>90%) in most categories except nervous system conditions.

The APD was highly sensitive in recording certain conditions among liveborn babies; for example, the sensitivity was 100% for gastroschisis and spina bifida. Conditions for which the APD had low sensitivity included cystic fibrosis (69%) and phenylketonuria (47%).

Overall 62% of audited medical records were eligible to be included on the RoCC, with a higher proportion (89%) for records of termination of pregnancy or stillbirth. Diagnoses of congenital conditions recorded on the APD were generally reliable, showing excellent agreement with RoCC diagnoses (Kappa > 0.9) for several important conditions including cystic fibrosis, trisomy 21, and spina bifida.

The strength of this study is that it is population based, covering all cases reported to the RoCC in 2009 and/or admitted to hospital at less than 1 year of age and eligible to be registered on the RoCC in 2009. The most important limitation of the study is that it did not include cases of congenital conditions that were diagnosed outside the hospital setting and were not reported to the RoCC. Also, as medical record review was not carried out for about 10% of unlinked APD records, the true sensitivity of the RoCC at a population level is likely to be lower than reported here.

We found that the quality and coverage of the RoCC depends on the condition of interest. While some congenital conditions are well reported on the RoCC, the APD has potential to substantially improve reporting of congenital conditions to the RoCC.

# 1. INTRODUCTION

The NSW Register of Congenital Conditions (RoCC)—formerly known as the NSW Birth Defects Register—is a statutory data collection under the *NSW Public Health Act 2010* (the Act). Under the Act, doctors, hospitals and laboratories are required to report certain congenital conditions detected during pregnancy or at birth or diagnosed in infants up to 1 year of age to the RoCC. These conditions include structural and chromosomal anomalies and the following genetic conditions: cystic fibrosis, phenylketonuria, congenital hypothyroidism and thalassaemia major. The RoCC assists in responding to apparent clusters of congenital conditions occurring in the community, identifying changes in incidence that may require investigation, and monitoring the occurrence of congenital conditions for service planning.

Congenital conditions are also recorded in NSW Admitted Patient Data (APD) for:

1. Mothers admitted to hospital for care of a fetus with a congenital condition.
2. Babies where the congenital condition was diagnosed at birth in hospital.
3. Infants admitted to hospital in the 1st year of life for care relating to a congenital condition.

Linkage of routinely collected administrative health data is a cost-effective and efficient technique to validate information contained in population health datasets. We carried out a quality assurance project using linked RoCC and APD records, and medical record review of unlinked APD records, to evaluate the enumeration of cases of congenital conditions reported to the RoCC, and to assess the quality of information on cases reported on the APD.

The aims of this project were to:

1. determine the number and type of confirmed congenital conditions reported to the APD and not to the RoCC for 2009;
2. assess the contribution of APD review in supplementing the RoCC;
3. determine the sensitivity of the RoCC in detecting cases using the total count of cases reported on the APD and validated by medical record review plus cases reported to the RoCC as the gold standard;
4. assess the reliability of coding of congenital conditions on the APD; and
5. assess the efficiency of the linkage and review process.

## 2. METHODS

### 2.1 Ethical approval

Ethical approval for the project was obtained from the NSW Population and Health Services Research Ethics Committee (Ethics Ref: 2012-03-378). Site specific assessments were carried out by Local Health Districts.

### 2.2 Data sources

The project used 2 data collections:

#### 1. NSW Register of Congenital Conditions

The RoCC is a population-based surveillance system established to monitor congenital conditions detected during pregnancy or at birth, or diagnosed in infants up to 1 year of age. The RoCC was established in 1990 and is administered by the Centre for Epidemiology and Evidence of the NSW Ministry of Health. Doctors, hospitals and laboratories are required, under the NSW Public Health Act 2010, to notify certain congenital conditions detected during pregnancy or in an infant up to 1 year of age.

There are 3 types of conditions that are reported to the RoCC:

- a) conditions that affect the growth, development and health of the baby that are present before birth, such as cleft lip, dislocated hip and problems with the development of the heart, lungs or other organs;
- b) conditions due to changes in the number of the baby's chromosomes, such as Down syndrome; and
- c) four conditions due to changes in the baby's inherited genetic information: cystic fibrosis, phenylketonuria, congenital hypothyroidism and thalassaemia major.

Children with congenital conditions are commonly treated at the 3 children's hospitals in NSW: The Children's Hospital at Westmead, Sydney Children's Hospital at Randwick, and John Hunter Children's Hospital. Medical records at these hospitals are regularly reviewed by RoCC staff and congenital conditions are therefore validated prior to being entered onto the RoCC database.

Congenital conditions are coded using the British Paediatric Association (BPA) Classification of Diseases:<sup>1</sup> Modified Classification for the NSW Register of Congenital Conditions. Disorders of function such as blindness and deafness are not captured in the RoCC. Conditions are classified as major if they are sufficient to qualify a case for registration, and minor if they are insufficient but should be reported when found in association with a major condition.

The RoCC includes records relating to all pregnancy outcomes, including spontaneous abortion, termination of pregnancy, stillbirth and livebirth. Reporting of spontaneous abortions to the RoCC is known to be incomplete as investigations that might reveal a congenital condition are only carried out for mothers with a history of repeat spontaneous abortions.

#### 2. NSW Admitted Patient Data

NSW Admitted Patient Data (APD) comprises a census of all admitted patient services provided by NSW public hospitals, public psychiatric hospitals, public multi-purpose services, private hospitals, and private day procedure centres. The APD covers demographic and episode-related data for every hospital separation in NSW.

Principal diagnosis and co-morbidities are coded according to the International Classification of Diseases and Related Health Problems, Tenth Revision, Australian Modification (ICD-10-AM), and procedures are coded according to the Australian Classification of Health Interventions (ACHI).<sup>2</sup>

### 2.3 Record selection for data linkage

The following records were selected:

- a) RoCC records with a registration year of 2009, where the outcome of pregnancy was livebirth, termination of pregnancy, stillbirth, or unknown.
- b) APD records:
  - i) records of babies born alive in the period 1 January to 31 December 2009 who had a hospital separation within 1 year of birth with a diagnosis–procedure code indicating a congenital condition eligible for inclusion on the RoCC. Codes include: ICD-10-AM codes listed in Appendix 2; or, an ICD-10-AM code beginning with Q, excluding those listed in Appendix 3; or, a procedure code for correction of talipes or undescended testis listed in Appendix 2; and
  - ii) records for women who were discharged from hospital following a medical abortion, termination of pregnancy–related procedure, or stillbirth in the period 1 January to 31 December 2009 with diagnosis codes indicating a fetus affected by a congenital condition (Appendix 1), and excluding any records where livebirth was an outcome.



## 2.4 Data linkage

Record linkage was carried out by the Centre for Health Record Linkage (CHeReL).<sup>3</sup> The CHeReL uses a privacy-preserving procedure whereby record linkage, which requires access to personal identifiers, is separated from the analysis of linked health records, which does not require access to personal identifiers. The CHeReL uses probabilistic record linkage methods and ChoiceMaker software.<sup>4,5</sup> ChoiceMaker uses 'blocking' and 'scoring' to identify definite and possible matches. During blocking, ChoiceMaker searches the target datasets for records that are possible matches to each other. There are 2 types of blocking. The exact blocking algorithm requires records to have the same set of valid fields and the same values for these fields. The automated blocking algorithm builds a set of conditions that are used to find as many as possible records that potentially match each other. Scoring employs a combination of a probabilistic decision, which is computed using a machine learning technique, and absolute rules, which include upper and lower probability cut-offs, to determine the final decision as to whether each potential match denotes or possibly denotes the same person. Indeterminate matches are reviewed manually.

Once the linkages were finalised, the CHeReL created a Project Person Number (PPN) for each person in the linked dataset, and assigned this PPN to the APD (Mothers–Babies) and RoCC (Mothers–Babies) records. The CHeReL returned the PPN and the encrypted record number from the source databases to the relevant data custodians. For this project, the rate of false positive links was reported to be zero, as all linked records with less than a perfect match probability were manually reviewed. The rate of missed links is estimated to be zero if full personal identifiers are available.

## 2.5 Medical record audit

The following records in the linked dataset were excluded from the audit of medical records:

- a) Medical records of the 3 children's hospitals were not audited, as they had been reviewed by RoCC staff prior to any record being included on the RoCC.
- b) Unlinked APD records for infants with a diagnosis code indicating talipes (club foot) and a procedure code indicating talipes repair were considered eligible for the RoCC without medical record audit (that is, automatically). Records with a diagnosis code indicating talipes without a relevant procedure code were considered ineligible for RoCC registration without audit.
- c) Unlinked APD records for infants with a diagnosis code indicating undescended testes and a procedure code associated with repair of undescended testes were considered eligible for registration on the RoCC without medical record audit (that is, automatically). Records with a diagnosis code indicating undescended testes without a relevant procedure code were considered ineligible for RoCC registration without audit.

Following these exclusions, a medical record review was carried out where a person (woman–infant) had an APD record but no linked RoCC record. The review was carried out by health information managers at relevant hospitals or by RoCC staff. Audit information was returned in de-identified format to the RoCC and entered onto a Microsoft Access database. Conditions were coded according to the BPA. Cases were categorised according to whether they were eligible to be registered on the RoCC or not eligible.

ICD-10-AM diagnosis codes that refer to minor conditions; that is, conditions that are only included when another major congenital condition is present, and non-contributory conditions are listed in Appendix 3.

## 2.6 Data preparation and cleaning

RoCC, APD and medical record audit data were merged into a single dataset. Further data preparation entailed:

- removing any redundant records, including records containing minor conditions only and APD mother records where there were multiple records for the same pregnancy;
- excluding APD records of non-NSW residents;
- flagging APD baby records where undescended testes or talipes was surgically corrected as automatically validated;
- consolidating APD congenital condition codes for each case onto 1 record where multiple records were present; and
- mapping RoCC BPA and ICD-10-AM codes to a common grouping for analysis.

## 2.7 Data analysis

All analyses were carried out using SAS v9.3.<sup>6</sup> Flowcharts for record selection, linkage and analysis for mothers and babies are shown in Figures 1 and 2 respectively.

### 2.7.1 Grouping of conditions

As information held on the RoCC is coded at a very fine level of detail, and many conditions are rare, congenital conditions were aggregated into the 14 *major diagnostic categories* used for reporting of congenital conditions in the *NSW Mothers and Babies* report, shown in Appendix 4.

For finer-grained analysis of conditions as coded on the RoCC and in the APD (for example, for calculating agreement), we also defined *specific condition groups* based on the finest grouping common to both BPA and ICD10-AM codes, with coarser groupings of rare conditions with related aetiology or site, also shown in Appendix 4.

### 2.7.2 Additional cases from APD

The number and type of confirmed additional cases found using APD data ( $n_{\text{additional confirmed}}$ ) was counted from distinct cases in the APD records that were not linked to an RoCC record, and that either had a corresponding audit record indicating that the case was eligible to be included on the RoCC, or were automatically deemed eligible because of a relevant procedure code (Figure 1—*mum\_apau\_elig*, Figure 2—*bub\_apau\_elig*, *bub\_ap\_ul\_auto*). Results were also stratified by major diagnostic category.

### 2.7.3 Sensitivity of the RoCC in case detection

The total number of records for each condition considered as the truth was calculated as the total number of infants reported as having the condition by the RoCC plus the number of valid APD records. The sensitivity of the RoCC in detecting true cases was calculated as the number of infants reported by the RoCC divided by the total, presented as a percentage. Ninety-five per cent binomial confidence intervals were calculated.

$$\text{sensitivity} = \frac{n_{\text{RoCC}}}{n_{\text{additional confirmed}} + n_{\text{RoCC}}} \quad (\text{as percentage})$$

$n_{\text{RoCC}}$  was derived from counts of distinct cases from *mum\_rc\_ppn* and *bub\_rc\_ppn* (see Figure 1 and Figure 2).

Cases where haemangioma or birth mark was the only major condition were excluded from the analysis. Results were also stratified by major diagnostic category. All records of talipes and undescended testes reported to the RoCC were included in the stratified analysis as major conditions, as major and minor cases could not be distinguished in the RoCC data.

The contribution of the APD to case finding was measured as  $(100 - \text{RoCC sensitivity}) \%$ .

### 2.7.4 Sensitivity of the APD

The total number of records for each condition considered as the truth was calculated as the total number of infants reported as having the condition by the RoCC plus the number of valid APD records. The sensitivity of the APD in detecting true cases was calculated as the number of infants reported by the APD divided by the total, presented as a percentage. Ninety-five per cent exact binomial confidence intervals were calculated.

$$\text{sensitivity} = \frac{n_{\text{APD}}}{n_{\text{additional confirmed}} + n_{\text{RoCC}}} \quad (\text{as percentage})$$

$n_{\text{APD}}$  was derived from counts of distinct cases from *mum\_ap\_ppn* and *bub\_ap\_ppn* (see Figure 1 and Figure 2).

Cases where haemangioma or birth mark was the only major condition were excluded from the analysis. Results were also stratified by major diagnostic category. All records of talipes or undescended testes in the RoCC were treated as major conditions for the purpose of the stratified analysis as major and minor cases could not be distinguished in the RoCC data.

## 2.7.5 Yield of APD record audit

The yield of the APD and medical record audit process was quantified by the ratio of confirmed additional cases found using APD data to the cases for which a medical record audit was completed. This reflects the efficiency of using the APD as a potential notification source in supplementing the RoCC.

Yield was calculated as follows:

$$\text{yield} = \frac{n_{\text{additional confirmed}}}{n_{\text{APD reviewed}}} \quad (\text{as percentage})$$

$n_{\text{APD reviewed}}$  was derived from counts of distinct cases from bub\_apau\_elig, bub\_apau\_inel, mum\_apau\_elig and mum\_apau\_inelig (see Figure 3 and Table 8).

Cases involving undescended testis or talipes were excluded from the analysis, as corrective surgery was not validated in the audit.

Confirmed additional cases were classified in two ways:

1. Within-category ascertainment ("Additional cases [strict]"), which is the number of confirmed additional cases that have the same major diagnostic category on the APD as that found in the medical record review.
2. Any-category ascertainment ("Additional cases [all]"), which is the number of confirmed additional cases regardless of whether the major diagnostic category on the APD was the same as that found in the medical record review.

Major diagnostic category-level yield calculations were performed for liveborn infants only.

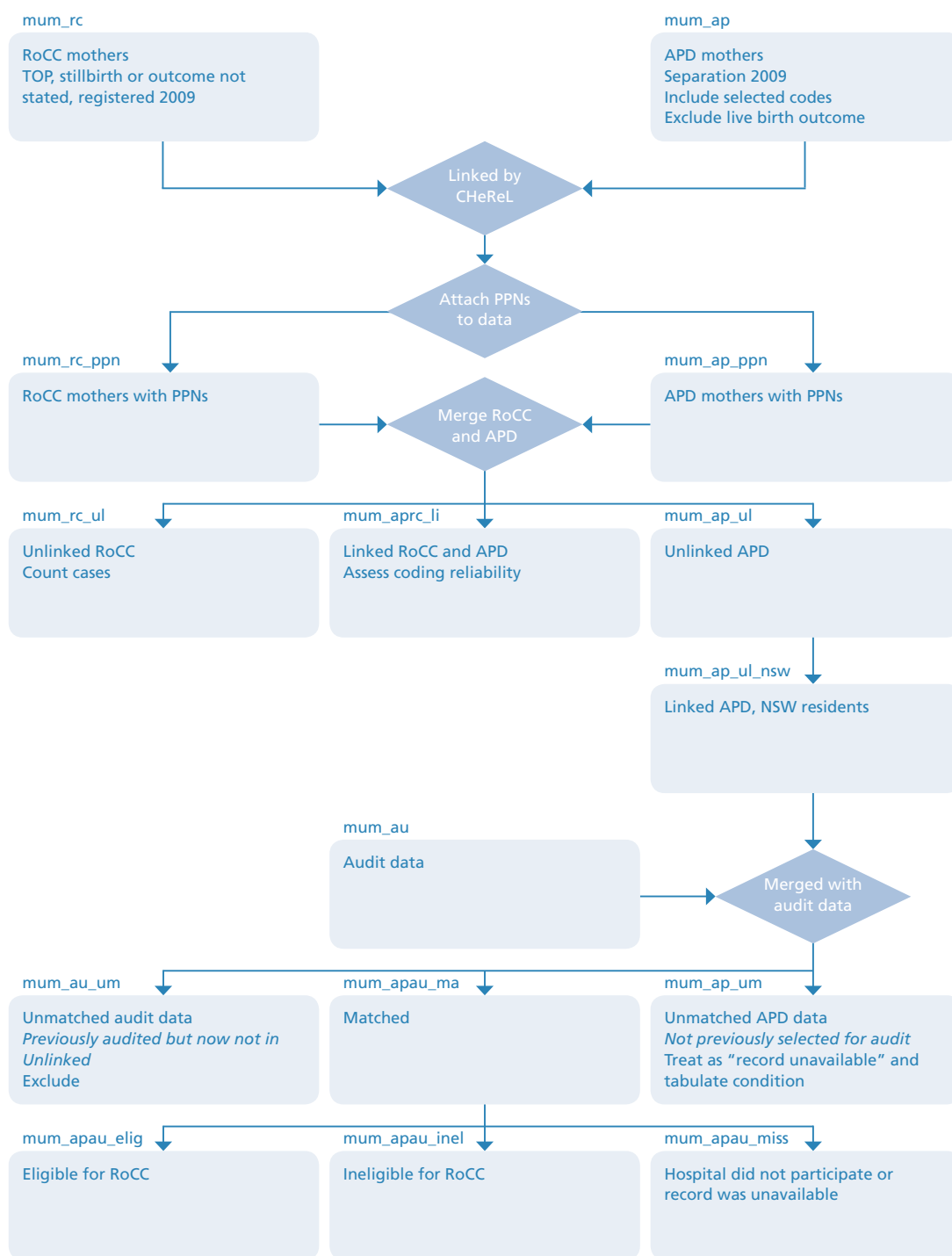
## 2.7.6 Agreement between APD and RoCC condition classification

Congenital condition codes were compared between the RoCC and matched APD records (Figure 1—mum\_aprc\_li; Figure 2—bub\_aprc\_li).

We calculated Cohen's Kappa for all specific condition groups that had 7 or more cases among linked RoCC cases. For each condition group we determined if a case had 1 or more condition codes falling into the condition group, according to APD diagnosis codes on any episode, and according to the RoCC record. All records of talipes or undescended testes in the APDC were treated as major conditions, as major and minor cases could not be distinguished in the RoCC data.

We also calculated the proportion of linked cases for which there was complete agreement in major conditions between RoCC and APD, based on specific condition groupings.

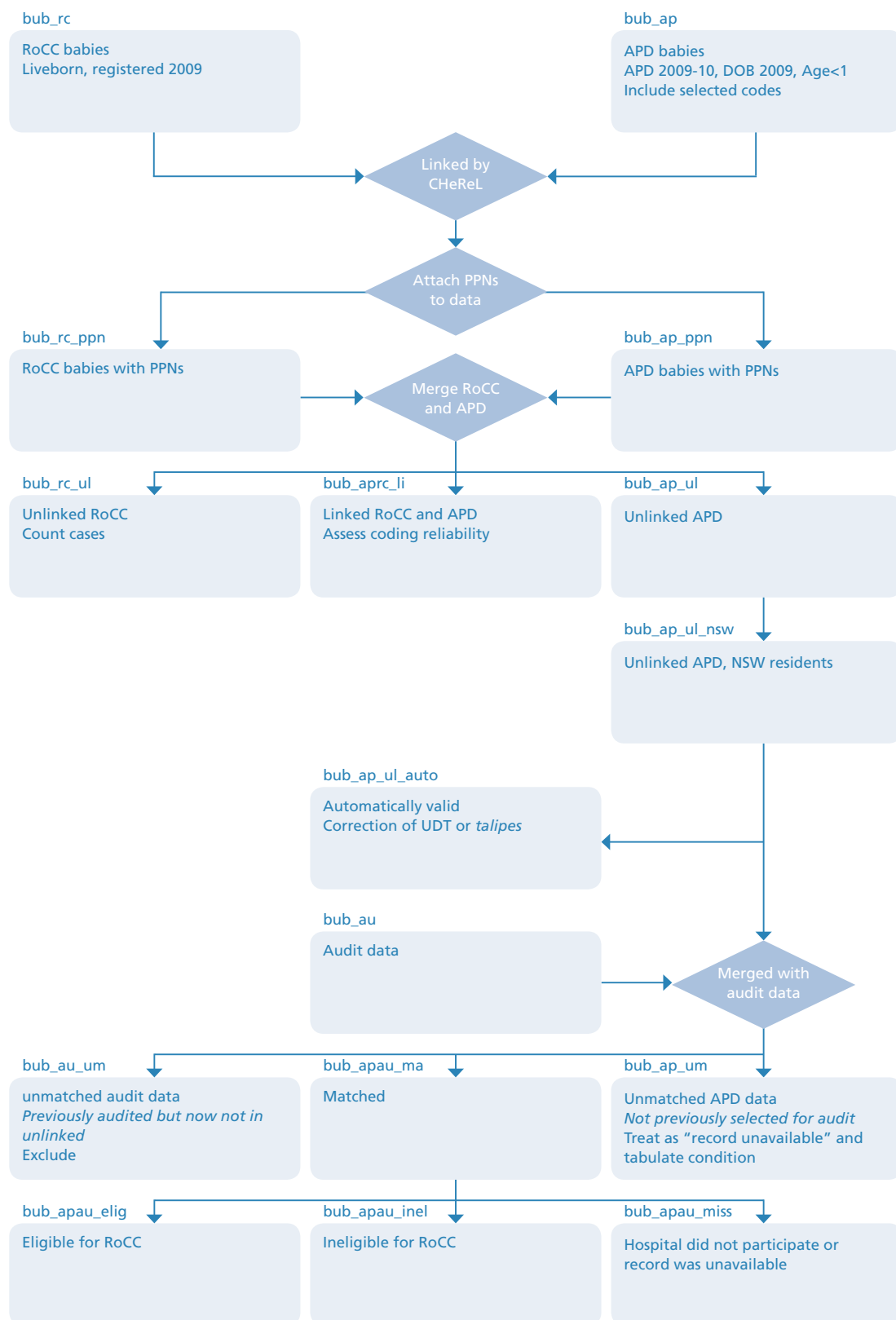
FIGURE 1: FLOWCHART OF RECORD SELECTION, LINKAGE AND ANALYSIS FOR MOTHERS



Note: Dataset names are printed above each box.

Source: NSW Register of Congenital Conditions (RoCC) 2009 and NSW Admitted Patient Data Collection (APD) 2009–2010, Centre for Epidemiology and Evidence, NSW Ministry of Health.

FIGURE 2: FLOWCHART OF RECORD SELECTION, LINKAGE AND ANALYSIS FOR BABIES



Note: Dataset names are printed above each box.

Source: NSW Register of Congenital Conditions (RoCC) 2009 and NSW Admitted Patient Data Collection (APD) 2009–2010, Centre for Epidemiology and Evidence, NSW Ministry of Health.

# 3. RESULTS

## 3.1 Record linkage and audit

A total of 7,586 records were selected for data linkage: 2,209 RoCC records and 5,377 APD records. The results of the record linkage are shown in Table 1.

**TABLE 1: RECORD LINKAGE RESULTS**

Data source	Linkage type	Results	
		Records No.	Persons No.
APD Women	APD records linked to RoCC records	359	347
	APD records not linked to RoCC records	159	157
RoCC Women	RoCC records linked to APD records	349	347
	RoCC records not linked to APD records	243	240
APD Babies	APD records linked to RoCC records	3,031	1,387
	APD records not linked to RoCC records	1,828	1,576
RoCC Babies	RoCC records linked to APD records	1,389	1387
	RoCC records not linked to APD records	228	228
<b>TOTAL</b>		<b>7,586</b>	<b>3935</b>

Source: NSW Register of Congenital Conditions (RoCC) 2009 and NSW Admitted Patient Data Collection (APD) 2009–2010, Centre for Epidemiology and Evidence, NSW Ministry of Health.

After data preparation, cleaning and restricting to NSW resident cases (Section 2.6) there were 1,776 unlinked APD records, representing 1,532 liveborn infants, and 152 unlinked APD records representing terminations of pregnancy or stillbirths (Table 2).

Of the total 1,547 records that qualified for audit, 1,378 records (89.1%) were reviewed, and had audit information complete and available for analysis (Table 3).

Of the total 90 hospitals with cases that qualified for audit, audits were carried out at 74 hospitals (82.2%). Sixty of 63 public hospitals (95.2%), and 13 of 27 private hospitals (48.1%) participated in the audit.

The disposition of unlinked NSW APD records and cases according to the merged audit data is shown in Table 3. Baby

records with a code for an undescended testes or talipes correction procedure were classified as automatically eligible for the RoCC. Cases were assigned to a single disposition category, and 'automatically eligible' took precedence over other categories.

Records for liveborn infants from The Children's Hospital at Westmead, Sydney Children's Hospital and John Hunter Hospital were not audited in this study. As part of routine quality assurance processes for the RoCC, all records from these hospitals with diagnosis codes associated with congenital conditions are audited, and therefore any eligible cases will already be present on the RoCC and not among unlinked APD data.

**TABLE 2: RECORDS AND CASES FOR ANALYSIS, NSW 2009**

Data source	Pregnancy outcome	Records No.	Cases No.	Linked cases No.	Unlinked records No.	Unlinked cases No.	Unlinked records NSW No.	Unlinked cases NSW No.
APD	Liveborn infant	4859	2963	1387	1828	1576	1776	1532
RoCC		1612	1612		225	225	225	225
APD	Termination of pregnancy, stillbirth or unknown	504	504	347	157	157	152	152
RoCC		590	590		243	243	243	243

Source: NSW Register of Congenital Conditions (RoCC) 2009 and NSW Admitted Patient Data Collection (APD) 2009–2010, Centre for Epidemiology and Evidence, NSW Ministry of Health.

**TABLE 3: UNLINKED APD DATA (NSW ONLY) BY PREGNANCY OUTCOME AND AUDIT DISPOSITION, NSW 2009**

Pregnancy outcome	Disposition	Records No.	Cases No.
Liveborn infant	Automatically eligible	97	96
	Audited – eligible for RoCC	765	657
	Audited – ineligible for RoCC	486	458
	Admitted to Children's hospital – ineligible for RoCC	284	201
	Not audited	144	120
	<b>TOTAL</b>	<b>1776</b>	<b>1532</b>
Termination of pregnancy, stillbirth or unknown	Audited – eligible for RoCC	113	113
	Audited – ineligible for RoCC	14	14
	Not audited	25	25
	<b>TOTAL</b>	<b>152</b>	<b>152</b>

Source: NSW Register of Congenital Conditions (RoCC) 2009 and NSW Admitted Patient Data Collection (APD) 2009–2010, Centre for Epidemiology and Evidence, NSW Ministry of Health.

### 3.2 Additional cases from APD

The medical record review of unlinked APD records confirmed a total of 866 additional cases of congenital conditions not recorded on the RoCC. Most additional cases among liveborn infants comprised conditions of the genitourinary system, musculoskeletal system and/or cardiovascular system, while most additional cases among terminations of pregnancy or stillbirths

comprised chromosomal abnormalities or conditions of the nervous system (Table 4). Additional nervous system conditions included 43 cases with neural tube defects: 1 liveborn case of encephalocele; among terminations of pregnancy, 4 cases of encephalocele, 30 cases of anencephaly and 5 cases of spina bifida; and 3 stillborn cases of anencephaly.

**TABLE 4: ADDITIONAL CASES OF CONGENITAL CONDITIONS REPORTED TO THE APD AND NOT TO THE RoCC BY PREGNANCY OUTCOME AND MAJOR DIAGNOSTIC CATEGORY, NSW 2009**

Pregnancy outcome–major diagnostic category	Cases No.	
<b>Live born</b>	Cardiovascular system	144
	Chromosomal anomalies	13
	Congenital malformation syndromes	7
	Ear, face and neck	2
	Eye	3
	Gastrointestinal system	13
	Genitourinary system	303
	Integumentary system	3
	Musculoskeletal system	253
	Nervous system	16
	Non-immune hydrops foetalis	1
	Other and unspecified anomalies	3
	Respiratory system	3
	Situs inversus	2
	<b>TOTAL*</b>	<b>753</b>
<b>Termination of pregnancy</b>	Cardiovascular system	2
	Chromosomal anomalies	49
	Congenital malformation syndromes	2
	Integumentary system	6
	Musculoskeletal system	5
	Nervous system	45
	Non-immune hydrops foetalis	5
	Other and unspecified anomalies	5
	<b>TOTAL*</b>	<b>109</b>

(Continued on next page)

TABLE 4: (Continued)

Pregnancy outcome–major diagnostic category		Cases No.
<b>Stillbirth</b>	Nervous system	4
	<b>TOTAL*</b>	<b>4</b>
<b>Total</b>	Cardiovascular system	146
	Chromosomal anomalies	62
	Congenital malformation syndromes	9
	Ear, face and neck	2
	Eye	3
	Gastrointestinal system	13
	Genitourinary system	303
	Integumentary system	9
	Musculoskeletal system	258
	Nervous system	65
	Non-immune hydrops foetalis	6
	Other and unspecified anomalies	8
	Respiratory system	3
	Situs inversus	2
	<b>TOTAL*</b>	<b>866</b>

\* As each case may have conditions falling into multiple major diagnostic categories, the total number of cases is less than the sum of cases in each category. Source: NSW Register of Congenital Conditions (RoCC) 2009 and NSW Admitted Patient Data Collection (APD) 2009–2010, Centre for Epidemiology and Evidence, NSW Ministry of Health.

### 3.3 Sensitivity of the RoCC and contribution of APD to case detection

Combining records of stillborn and liveborn infants and fetuses associated with terminations of pregnancy, along with cases with unknown outcome, the total number of infants and fetuses reported by the RoCC as having a congenital condition was 2,187 (excluding cases where haemangioma or birth mark was the only major condition). Addition of the 866 cases identified from the APD and verified by medical record audit gave a total of 3,053 cases of congenital conditions in 2009. The overall sensitivity of the RoCC was 71.6% (95% CI 70.0–73.2). The sensitivity of the RoCC was 68.0% for live born infants, and 83.9% for fetuses associated with termination of pregnancy, stillbirth or unknown pregnancy outcome.

The sensitivity of the RoCC by major diagnostic category and pregnancy outcome is shown in Table 5. There is a great deal of variation in sensitivities, from 54.8% in ascertainment of conditions of the genitourinary system in live born infants to over 95% for gastrointestinal system conditions. Counts of additional APD cases and RoCC cases are also shown in Figure 3.

Additional key results regarding the sensitivity of the RoCC in detecting congenital conditions within major diagnostic categories were:

- for the nervous system, the sensitivity of the RoCC in ascertaining anencephaly is low at 28.3%, due to substantial under-reporting among terminations of pregnancy (data not shown)
- for the cardiovascular system, the sensitivity of the majority of specific condition groups were above 90%; the lowest sensitivities were for ventricular septal defect (68.6%), atrial septal defect (77.4%) and stenosis of the pulmonary artery (69.2%)
- for the genitourinary system, sensitivities were less than 80% for many condition groups
- for the musculoskeletal system, sensitivities were above 80% for most condition groups, and low for unstable hip (26.0%), congenital dislocation of the hips (56.8%) and for polydactyly (66.0%).

The contribution of the APD to case detection varied substantially with the condition of interest. Inclusion of additional cases from the APD increased the enumeration of major diagnostic categories of the genitourinary system, musculoskeletal system and nervous system by 25% or more (Table 5). The APD contributed 35.8% to the total cases of neural tube defects.



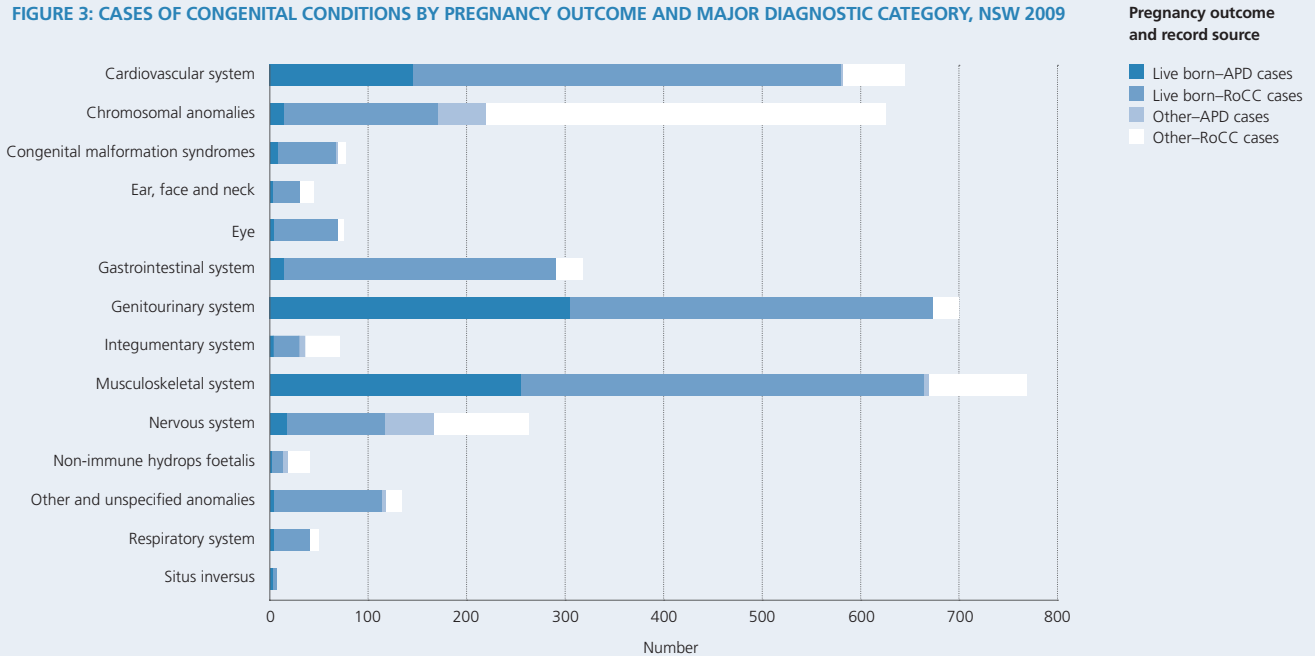
**TABLE 5: SENSITIVITY OF THE ROCC BY PREGNANCY OUTCOME AND MAJOR DIAGNOSTIC CATEGORY, NSW 2009**

Pregnancy outcome–major diagnostic category		RoCC Cases No.	Total Cases No.	Sensitivity %	95% CI
<b>Live born</b>	Cardiovascular system	433	577	75.0	71.3–78.5
	Chromosomal anomalies	156	169	92.3	87.2–95.8
	Congenital malformation syndromes	59	66	89.4	79.4–95.6
	Ear, face and neck	27	29	93.1	77.2–99.2
	Eye	65	68	95.6	87.6–99.1
	Gastrointestinal system	276	289	95.5	92.4–97.6
	Genitourinary system	367	670	54.8	50.9–58.6
	Integumentary system	26	29	89.7	72.7–97.8
	Musculoskeletal system	408	661	61.7	57.9–65.5
	Nervous system	100	116	86.2	78.6–91.9
	Non-immune hydrops foetalis	11	12	91.7	61.5–99.8
	Other and unspecified anomalies	109	112	97.3	92.4–99.4
	Respiratory system	37	40	92.5	79.6–98.4
	Situs inversus	4	6	66.7	22.3–95.7
	<b>TOTAL*</b>	<b>1597</b>	<b>2350</b>	<b>68.0</b>	<b>66.0–69.8</b>
<b>Termination of pregnancy, stillbirth or unknown</b>	Cardiovascular system	63	65	96.9	89.3–99.6
	Chromosomal anomalies	404	453	89.2	86.0–91.9
	Congenital malformation syndromes	8	10	80.0	44.4–97.5
	Ear, face and neck	15	15	100.0	78.2–100.0
	Eye	6	6	100.0	54.1–100.0
	Gastrointestinal system	27	27	100.0	87.2–100.0
	Genitourinary system	27	27	100.0	87.2–100.0
	Integumentary system	35	41	85.4	70.8–94.4
	Musculoskeletal system	99	104	95.2	89.1–98.4
	Nervous system	96	145	66.2	57.9–73.9
	Non-immune hydrops foetalis	23	28	82.1	63.1–93.9
	Other and unspecified anomalies	16	21	76.2	52.8–91.8
	Respiratory system	9	9	100.0	66.4–100.0
	<b>TOTAL*</b>	<b>590</b>	<b>703</b>	<b>83.9</b>	<b>81.0–86.6</b>
	<b>Total</b>	Cardiovascular system	496	642	77.3
Chromosomal anomalies		560	622	90.0	87.4–92.3
Congenital malformation syndromes		67	76	88.2	78.7–94.4
Ear, face and neck		42	44	95.5	84.5–99.4
Eye		71	74	96.0	88.6–99.2
Gastrointestinal system		303	316	95.9	93.1–97.8
Genitourinary system		394	697	56.5	52.8–60.3
Integumentary system		61	70	87.1	77.0–94.0
Musculoskeletal system		507	765	66.3	62.8–69.6
Nervous system		196	261	75.1	69.4–80.2
Non-immune hydrops foetalis		34	40	85.0	70.2–94.3
Other and unspecified anomalies		125	133	94.0	88.5–97.4
Respiratory system		46	49	93.9	83.1–98.7
Situs inversus		4	6	66.7	22.3–95.7
<b>TOTAL*</b>		<b>2187</b>	<b>3053</b>	<b>71.6</b>	<b>70.0–73.2</b>

\* As each case may have conditions falling into multiple major diagnostic categories, the total number of cases is less than the sum of cases in each category.

Source: NSW Register of Congenital Conditions (RoCC) 2009 and NSW Admitted Patient Data Collection (APD) 2009–2010, Centre for Epidemiology and Evidence, NSW Ministry of Health.

**FIGURE 3: CASES OF CONGENITAL CONDITIONS BY PREGNANCY OUTCOME AND MAJOR DIAGNOSTIC CATEGORY, NSW 2009**



Source: NSW Register of Congenital Conditions (RoCC) 2009 and NSW Admitted Patient Data Collection (APD) 2009–2010, Centre for Epidemiology and Evidence, NSW Ministry of Health.

Note: Counts of APD cases comprise additional APD cases only.

### 3.4 Sensitivity of the APD

Combining records of stillborn and liveborn infants and fetuses associated with terminations of pregnancy, the total number of infants and fetuses found on the APD and either confirmed by audit or linkage to RoCC cases was 2,595.

The overall sensitivity of the APD was 85.0% (95% CI 83.7–86.3). The sensitivity of the APD was 90.9% for live born infants, and 65.4% for fetuses associated with termination of pregnancy, stillbirth or unknown pregnancy outcome.

The sensitivity of the APD by major diagnostic category and pregnancy outcome is shown in Table 6. There was a great deal of variation in sensitivities, from 18.5% in

ascertainment of conditions of the genitourinary system in termination of pregnancy, stillbirth or unknown pregnancy outcome to over 90% for gastrointestinal system conditions. For the nervous system, the sensitivity of the APD was high for anencephaly (100%), spina bifida (94.8%) and encephalocele (81.3%). For the musculoskeletal system, the sensitivity was variable but high for gastroschisis (97.1%). For the cardiovascular system, the sensitivity for many of the specific condition groups was above 90%; the lowest sensitivity was for hypoplastic left ventricle (30.8%). For the genitourinary system, sensitivities were greater than 80% for most condition groups.

**TABLE 6: SENSITIVITY OF THE APD BY PREGNANCY OUTCOME AND MAJOR DIAGNOSTIC CATEGORY, NSW 2009**

Pregnancy outcome–major diagnostic category		APD cases No.	Total cases No.	Sensitivity %	95% CI
<b>Live born infant</b>	Cardiovascular system	556	577	96.4	94.5–97.7
	Chromosomal anomalies	143	169	84.6	78.3–89.7
	Congenital malformation syndromes	64	66	97.0	89.5–99.6
	Ear, face and neck	24	29	82.8	64.2–94.2
	Eye	66	68	97.1	89.8–99.6
	Gastrointestinal system	286	289	99.0	97.0–99.8
	Genitourinary system	636	670	94.9	93.0–96.5
	Integumentary system	23	29	79.3	60.3–92.0
	Musculoskeletal system	606	661	91.7	89.3–93.7
	Nervous system	110	116	94.8	89.1–98.1
	Non-immune hydrops foetalis	10	12	83.3	51.6–97.9
	Other and unspecified anomalies	53	112	47.3	37.8–57.0
	Respiratory system	36	40	90.0	76.3–97.2
	Situs inversus	6	6	100.0	54.1–100.0
	<b>TOTAL*</b>	<b>2135</b>	<b>2350</b>	<b>90.9</b>	<b>89.6–92.0</b>
<b>Termination of pregnancy, stillbirth or unknown</b>	Cardiovascular system	22	65	33.9	22.6–46.7
	Chromosomal anomalies	328	453	72.4	68.0–76.5
	Congenital malformation syndromes	3	10	30.0	6.7–65.3
	Ear, face and neck	6	15	40.0	16.3–67.7
	Eye	2	6	33.3	4.3–77.7
	Gastrointestinal system	9	27	33.3	16.5–54.0
	Genitourinary system	5	27	18.5	6.3–38.1
	Integumentary system	26	41	63.4	46.9–77.9
	Musculoskeletal system	31	104	29.8	21.2–39.6
	Nervous system	109	145	75.2	67.3–82.0
	Non-immune hydrops foetalis	19	28	67.9	47.7–84.1
	Other and unspecified anomalies	10	21	47.6	25.7–70.2
	Respiratory system	3	9	33.3	7.5–70.1
	<b>TOTAL*</b>	<b>460</b>	<b>703</b>	<b>65.4</b>	<b>61.8–69.0</b>
	<b>Total</b>	Cardiovascular system	578	642	90.0
Chromosomal anomalies		471	622	75.7	72.2–79.0
Congenital malformation syndromes		67	76	88.2	78.7–94.4
Ear, face and neck		30	44	68.2	52.4–81.4
Eye		68	74	91.9	83.2–97.0
Gastrointestinal system		295	316	93.4	90.0–95.8
Genitourinary system		641	697	92.0	89.7–93.9
Integumentary system		49	70	70.0	57.9–80.4
Musculoskeletal system		637	765	83.3	80.4–85.9
Nervous system		219	261	83.9	78.9–88.2
Non-immune hydrops foetalis		29	40	72.5	56.1–85.4
Other and unspecified anomalies		63	133	47.4	38.7–56.2
Respiratory system		39	49	79.6	65.7–89.8
Situs inversus		6	6	100.0	54.1–100.0
<b>TOTAL*</b>		<b>2595</b>	<b>3053</b>	<b>85.0</b>	<b>83.7–86.3</b>

\* As each case may have conditions falling into multiple major diagnostic categories, the total number of cases is less than the sum of cases in each category.

Source: NSW Register of Congenital Conditions (RoCC) 2009 and NSW Admitted Patient Data Collection (APD) 2009–2010, Centre for Epidemiology and Evidence, NSW Ministry of Health.

### 3.5 Yield of APD record audit

Medical record audits were completed for total of 1,242 unlinked cases from the APD, of which 770 (62.0%) were identified as having a congenital condition that was eligible to be registered on the RoCC (Table 7). Audit of records for live born babies had a lower yield (58.9%) than

audit of records for termination of pregnancy or stillbirth (89.0%). Analysing yield by major diagnostic category for live born babies showed some variation. Yields were low for conditions of the eye (19.1%), gastrointestinal system (3.0%) and integumentary system (16.7%) (Table 7).

**TABLE 7: YIELD OF APD RECORD AUDIT BY PREGNANCY OUTCOME AND MAJOR DIAGNOSTIC CATEGORY, NSW 2009**

Pregnancy outcome–major diagnostic category		Additional cases (strict) No.	Additional cases (all) No.	Audited cases No.	Yield (all)	
					%	95% CI
<b>Live born baby</b>	Cardiovascular system	139	141	208	67.8	61.0–74.1
	Chromosomal anomalies	12	12	14	85.7	57.2–98.2
	Congenital malformation syndromes	7	10	15	66.7	38.4–88.2
	Ear, face and neck	2	3	16	18.8	4.1–45.7
	Eye	3	4	21	19.1	5.5–41.9
	Gastrointestinal system	12	18	60	30.0	18.9–43.2
	Genitourinary system	214	222	345	64.4	59.0–69.4
	Integumentary system	3	4	24	16.7	4.7–37.4
	Musculoskeletal system	240	242	388	62.4	57.3–67.2
	Nervous system	15	17	36	47.2	30.4–64.5
	Non-immune hydrops foetalis	1	1	2	50.0	1.3–98.7
	Other and unspecified anomalies	3	5	20	25.0	8.7–49.1
	Respiratory system	2	2	10	20.0	2.5–55.6
	Situs inversus	2	2	2	100.0	15.8–100.0
	<b>TOTAL*</b>		<b>657</b>	<b>1115</b>	<b>58.9</b>	<b>56.0–61.8</b>
<b>Termination of pregnancy, stillbirth or unknown</b>			113	127	89.0	82.2–93.8
<b>TOTAL</b>			<b>770</b>	<b>1242</b>	<b>62.0</b>	<b>59.2–64.7</b>

\* As each case may have conditions falling into multiple major diagnostic categories, the total number of cases is less than the sum of cases in each category.

Source: NSW Register of Congenital Conditions (RoCC) 2009 and NSW Admitted Patient Data Collection (APD) 2009–2010, Centre for Epidemiology and Evidence, NSW Ministry of Health.

### 3.6 Agreement between APD and RoCC condition classification

Of the 78 specific condition groupings for which we calculated a kappa value, kappa was >0.7 for 46. Cystic fibrosis, phenylketonuria, trisomy 21, gastroschisis and spina bifida were among the groups for which kappa was >0.9 (Table 8). Note that these calculations were among cases that linked between RoCC and APD (i.e. excluding most APD false positives), and might not be representative

of agreement rates in an unselected population of live born babies.

The agreement according to the more exact definition of a complete match on each case’s major conditions (based on specific condition groupings) was 53.1% (95% CI 50.4–55.7).

**TABLE 8: AGREEMENT BETWEEN APD AND RoCC CONDITION CLASSIFICATIONS IN LINKED CASES, NSW 2009**

Condition	Kappa	95% CI	No.
Phenylketonuria	1.000	1.000–1.000	7
Cystic fibrosis	1.000	1.000–1.000	19
Trisomy 21	0.992	0.978–1.000	70
Gastroschisis	0.983	0.950–1.000	31
Hypospadias	0.963	0.937–0.988	118
Diaphragmatic hernia	0.962	0.910–1.000	27
Exomphalos	0.956	0.895–1.000	22
Spina Bifida	0.952	0.885–1.000	20
Congenital cataract	0.952	0.885–1.000	21
Hirschsprungs Disease	0.944	0.866–1.000	17
Common arterial trunk	0.941	0.825–1.000	8
Atresia of hepatic or bile ducts	0.941	0.825–1.000	8
Horseshoe kidney	0.933	0.802–1.000	8
Total or partial anomalous pulmonary venous return	0.928	0.828–1.000	15
Pierre Robin syndrome	0.922	0.815–1.000	12
Polydactyly	0.914	0.858–0.970	55
Cystic kidney disease	0.908	0.818–0.998	20
Oesophageal atresia with TOF	0.903	0.809–0.997	19
Atresia-stenosis of small intestine (includes with fistula)	0.903	0.809–0.998	20
Choanal atresia and stenosis	0.899	0.760–1.000	10
Transposition of great vessels	0.895	0.833–0.956	53
Craniosynostosis	0.891	0.804–0.977	28
Buphthalmos-congenital glaucoma	0.888	0.734–1.000	9
Hydrops not due to isoimmunisation	0.888	0.734–1.000	9
Atresia-stenosis of anus (includes with fistula)	0.877	0.797–0.956	35
Posterior urethral valves	0.874	0.702–1.000	8
Cleft lip and palate	0.861	0.787–0.936	50
Atrioventricular septal defect	0.860	0.758–0.963	23
Tetralogy of Fallot	0.853	0.771–0.935	37
Duplex kidney and/or collecting system	0.849	0.738–0.959	26
Renal agenesis – dysgenesis	0.843	0.736–0.950	27
Undescended testis	0.833	0.767–0.898	77
Ventricular septal defect	0.824	0.779–0.869	164
Sex chromosome anomalies	0.799	0.605–0.992	11
Talipes	0.794	0.693–0.896	39
Coarctation of aorta	0.789	0.694–0.884	48
Reduction deformity of corpus callosum	0.784	0.615–0.952	16
Cleft palate only	0.755	0.672–0.838	58
Reduction deformities of limbs	0.753	0.629–0.878	31
Cleft lip only	0.730	0.617–0.843	29
Hydronephrosis	0.726	0.635–0.816	65
Dextrocardia	0.725	0.514–0.936	10
Congenital dislocation of hips	0.722	0.618–0.826	49
Congenital hydrocephalus	0.719	0.572–0.865	27
Anomalies of ribs and sternum	0.718	0.517–0.918	15
Hypoplastic heart	0.703	0.556–0.851	30
Atresia of pulmonary valve	0.698	0.468–0.928	8
Microcephaly	0.691	0.537–0.844	25
Bicuspid aortic valve	0.689	0.539–0.838	8
Autosomal chromosome deletions	0.689	0.539–0.838	30
Anophthalmos-microphthalmos	0.686	0.492–0.881	17
Stenosis of pulmonary valve	0.680	0.548–0.813	34
Pelvic-ureteric junction obstruction	0.679	0.549–0.808	29

(Continued on next page)

**TABLE 8: (Continued)**

Condition	Kappa	95% CI	No.
Congenital hypothyroidism	0.664	0.458–0.870	9
Anomalies of spinal vertebrae	0.617	0.409–0.825	19
Hypoplasia of optic nerve	0.614	0.302–0.925	9
Indeterminate sex - ambiguous genitalia	0.614	0.302–0.926	8
Aplasia or hypoplasia of testes or scrotum	0.614	0.301–0.926	7
Anomalies of vitreous humour	0.599	0.239–0.958	7
Atrial septal defect	0.598	0.535–0.661	196
Unstable hip	0.590	0.463–0.717	38
Atresia of pulmonary artery	0.586	0.307–0.865	7
Ectopic anus	0.569	0.315–0.822	15
Stenosis of aortic valve	0.531	0.224–0.838	8
Syndactyly	0.496	0.227–0.766	11
Atresia of tricuspid valve	0.468	0.174–0.762	12
Common ventricle	0.467	0.173–0.762	9
Osteodystrophy	0.459	0.123–0.796	4
Hydroureter and megaloureter	0.381	0.178–0.585	23
Coloboma of iris	0.331	-0.014–0.676	4
Intestinal malrotation	0.313	0.042–0.583	15
Stenosis of pulmonary artery	0.253	0.067–0.439	17
Atresia of aortic valve	0.220	-0.141–0.582	7
Nose, larynx, trachea, bronchus	0.216	0.036–0.396	7
Adenomatoid malformation of lung	0.153	-0.112–0.418	12
Tracheo-oesophageal fistula (TOF) only	0.139	-0.111–0.389	5
Ureterocoele	-0.004	-0.007–0.001	11

Source: NSW Register of Congenital Conditions (RoCC) 2009 and NSW Admitted Patient Data Collection (APD) 2009–2010, Centre for Epidemiology and Evidence, NSW Ministry of Health.

## 4. Discussion

We carried out a quality assurance project by linking RoCC and APD records for 2009 and reviewing medical records of unlinked APD records. We evaluated the enumeration of cases of congenital conditions reported to the RoCC and assessed the quality of information on cases reported on the APD.

There were 2,187 cases of congenital conditions reported to the RoCC in 2009. The medical record review of unlinked APD records confirmed a total of 866 additional cases of congenital conditions that were not recorded on the RoCC, giving a total of 3,053 cases of congenital conditions for 2009. The additional cases comprised 753 (87.0%) livebirths, 109 (12.6%) terminations of pregnancy and 4 (0.5%) stillbirths.

The contribution of the APD to the total case enumeration was 28.4%. The additional cases were most commonly associated with genitourinary or musculoskeletal conditions (babies) and chromosomal abnormalities or nervous system conditions (mothers). In relation to neural tube defects, 77 cases were reported to the RoCC while the medical record audit revealed an additional 43 cases, giving a total of 120 cases for 2009. The additional cases comprised 1 livebirth, 3 stillbirths and 39 terminations of pregnancy.

The overall sensitivity of the RoCC was 71.6%. The RoCC was highly sensitive (>90%) for chromosomal abnormalities, eye conditions, gastrointestinal conditions and respiratory conditions among liveborn babies. Among terminations of pregnancy, stillbirths and cases with unknown outcome the RoCC was highly sensitive (>90%) in most categories except nervous system conditions.

Coding of congenital conditions on the APD for live born babies was variable but highly sensitive for some conditions, e.g. sensitivity of the APD was 100% (33/33) for gastroschisis and 100% (20/20) for spina bifida. Conditions for which APD coding was poorly sensitive included cystic fibrosis 69% (20/29) and phenylketonuria 47% (7/15). The lower sensitivity of the APD for congenital conditions in cases with termination of pregnancy or stillbirth (65%) suggests underuse of the ICD10-AM codes for *maternal care for known or suspected fetal abnormality and damage*.

The yield of the medical record review was high (overall 62%), especially for records of termination of pregnancy or stillbirth (89%), indicating a low false-positive rate. The variation in yield by diagnostic category is plausible given that there is often only a subtle distinction between certain RoCC eligible and ineligible conditions, and ICD10-AM codes may not capture these differences well.

Among cases that could be linked between RoCC and APD, agreement between the ICD10-AM diagnosis codes and BPA classification codes was variable but generally good. There was excellent agreement (Kappa > 0.9) for several important conditions such as cystic fibrosis, trisomy 21 and spina bifida.

Ascertainment of talipes and undescended testis via APD procedure codes appeared reliable based on kappa (>0.7); sensitivity for talipes was moderate (69%) and for undescended testis was excellent (98%). The sensitivity findings are conservative as any RoCC minor cases were treated as major for the purpose of the analysis.

The strength of this study is that it is population based, covering all cases reported to the RoCC in 2009 and/or admitted to hospital at less than 1 year of age and eligible to be registered on the RoCC in 2009. We found substantial under-reporting to the RoCC of congenital condition cases admitted to hospital, while demonstrating that hospital data had good sensitivity, low false positive rates, and agreed well with the RoCC for reported cases.

Limitations of the study include:

1. We were unable to ascertain the number of cases of congenital conditions that were diagnosed outside the hospital setting and were not reported to the RoCC. Also, medical record review was not carried out for about 10% of unlinked APD records. The true sensitivities of the RoCC and APD at a population level are therefore likely to be lower than reported here.
2. Some RoCC records with an unknown pregnancy outcome of the (88 cases) in all for 2009 may represent a live born baby that was not reported to the RoCC at birth. Any such births in the APD data would be present as baby records, and in this study could not be linked to their corresponding RoCC records. These false negative linkages would artificially raise the count of additional cases of congenital conditions, resulting in a decrease in apparent RoCC sensitivity, an increase in the apparent contribution of the linkage and audit process to known cases, and an increase in the apparent yield of the audit process. In the worst case, this would decrease the apparent sensitivity of the RoCC by about 2.5 percentage points.
3. Agreement was calculated using specific condition groupings that were chosen for convenience and as the finest grouping common to the disease classifications used by both the RoCC and the APD. These groupings were often broader than the codes used by the RoCC, limiting inferences that we can make about the contribution of the APD to enumeration of specific conditions.

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4. Disagreement between the RoCC and APD may stem from the different intentions behind recording a condition in each system. The RoCC records any condition diagnosed, while diagnoses in the APD represent conditions requiring clinical care at the time of hospital stay. APD diagnoses may also describe the consequences of congenital conditions rather than the primary condition itself, or describe the individual's state after corrective treatment. For example, in the case of ureterocoele recorded on the RoCC, the corresponding APD diagnosis or diagnoses may be obstructive defects of renal pelvis and ureter, megaloureter, hydroureter or hydronephrosis, depending on the course of the disease.
5. Although the false positive and false negative rates for the linkage process were low for this study, the quality of linkage is likely to have varied by pregnancy outcome. A study of NSW for data for 2001–08 found approximately 4% of registered stillbirths could not be linked to a maternal hospital inpatient record, compared with less than 1% of registered livebirths.<sup>7</sup> Missed links would affect the results as described in 2. above.
6. Some RoCC cases that did not link to the APD may represent a spontaneous abortion or a birth or termination outside NSW, data for which were out of scope for this project.

linkage could be considered. This would provide a well-defined frame for analysis of liveborn and stillborn babies, allowing complete enumeration of true negative cases and estimation of ascertainment specificity.

The results of this study indicate that a substantial number of cases of congenital conditions are currently reported to the APD and not to the RoCC. Automated reporting of congenital conditions from the APD without medical record review could be considered for specific conditions where agreement between RoCC and APD is high, and APD sensitivity and yield are high, e.g. gastroschisis and spina bifida. For other conditions reported on the APD, case registration on the RoCC would require a notification process and medical record review, similar to that already carried out by the RoCC to validate new notifications. Alternatively, the APD could be used to assess case ascertainment on the RoCC using record linkage; and, should a systematic under-ascertainment of certain types of congenital conditions be found prospectively, the APD cases that have not been notified could be followed up with the relevant hospital.

Future work could take advantage of the CHeReL family linkage process, in which parent–child relationships give a greater chance of correct linkage. Family linkage has the added benefit of identifying twins in the hospital data and allowing some types of misclassification in the RoCC to be resolved (such as live born babies registered as 'unknown' outcome, described above).<sup>7</sup> Inclusion of the NSW perinatal data collection in the



## 5. Conclusion

This study found that a substantial number of cases of congenital conditions are currently reported to the APD and not to the RoCC. The medical record review of APD records confirmed a total of 866 additional cases of congenital conditions in 2009 that were not recorded on the RoCC, most of which related to liveborn babies. The contribution of the APD to the total case enumeration for 2009 was 28.4%. On the other hand, the RoCC was highly sensitive (>90%) for chromosomal abnormalities, eye conditions, gastrointestinal conditions and respiratory conditions among liveborn babies.

Diagnoses of congenital conditions recorded on the APD were generally reliable. The sensitivity of the APD was 90.9% for liveborn babies. Agreement between the APD and RoCC diagnoses was generally good, with excellent agreement (Kappa > 0.9) for several important conditions such as cystic fibrosis, trisomy 21 and spina bifida. Medical record review found 62% of APD cases were eligible to be included on the RoCC.

The APD has significant potential for enhancing the reporting of congenital conditions to the RoCC. This could take the form of automated reporting of specific congenital conditions that have demonstrated good agreement with the RoCC, or a general notification and medical record review process. Alternatively, ongoing data linkage could identify under-ascertainment to be followed up with the relevant hospital.

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# 7. APPENDICES

## Appendix 1: ACHI and ICD-10-AM codes for mother inclusion and exclusion criteria

Code	Description
<i>Termination of pregnancy procedure codes (ACHI)</i>	
35640-00	Dilation and curettage of uterus
35640-01	Curettage of uterus without dilation
35640-03	Suction curettage of uterus
35643-03	Dilation and evacuation of uterus (2nd trimester)
90461-00	Intra-amniotic injection for abortion
90462-00	Insertion of prostaglandin suppository for induction of abortion
90463-00	Fetal reduction
90465-00	Medical induction of labour, oxytocin
90465-01	Medical induction of labour, prostaglandin
90465-02	Other medical induction of labour
90465-05	Medical and surgical induction of labour
<i>Termination of pregnancy diagnosis codes (ICD-10-AM)</i>	
O04	Medical abortion
O04.0	Medical abortion - incomplete, complicated by infection
O04.1	Medical abortion - incomplete, complicated by haemorrhage
O04.2	Medical abortion - incomplete, complicated by embolism
O04.3	Medical abortion - incomplete, with other/unspecified complications
O04.4	Medical abortion - incomplete, without complication
O04.5	Medical abortion - complete or unspecified, complicated by infection
O04.6	Medical abortion - complete or unspecified, complicated by haemorrhage
O04.7	Medical abortion - complete or unspecified, complicated by embolism
O04.8	Medical abortion - complete or unspecified, with other/unspecified complications
O04.9	Medical abortion - complete or unspecified, without complication
O05	Other abortion
O05.0	Other abortion - incomplete, complicated by infection
O05.1	Other abortion - incomplete, complicated by haemorrhage
O05.2	Other abortion - incomplete, complicated by embolism
O05.3	Other abortion - incomplete, with other/unspecified complications
O05.4	Other abortion - incomplete, without complication
O05.5	Other abortion - complete or unspecified, complicated by infection
O05.6	Other abortion - complete or unspecified, complicated by haemorrhage
O05.7	Other abortion - complete or unspecified, complicated by embolism
O05.8	Other abortion - complete or unspecified, with other/unspecified complications
O05.9	Other abortion - complete or unspecified, without complication
O06	Unspecified abortion
O06.0	Unspecified abortion - incomplete, complicated by infection
O06.1	Unspecified abortion - incomplete, complicated by haemorrhage
O06.2	Unspecified abortion - incomplete, complicated by embolism
O06.3	Unspecified abortion - incomplete, with other/unspecified complications
O06.4	Unspecified abortion - incomplete, without complication
O06.5	Unspecified abortion - complete or unspecified, complicated by infection
O06.6	Unspecified abortion - complete or unspecified, complicated by haemorrhage
O06.7	Unspecified abortion - complete or unspecified, complicated by embolism
O06.8	Unspecified abortion - complete or unspecified, with other/unspecified complications
O06.9	Unspecified abortion - complete or unspecified, without complication
O07.0	Failed medical abortion, complicated by genital tract and pelvic infection
O07.1	Failed medical abortion, complicated by delayed or excessive haemorrhage
O07.2	Failed medical abortion, complicated by embolism
O07.3	Failed medical abortion, with other and unspecified complications
O07.4	Failed medical abortion, without complication
O07.5	Other and unspecified failed attempted abortion, complicated by genital tract and pelvic infection
O07.6	Other and unspecified failed attempted abortion, complicated by delayed or excessive haemorrhage
O07.7	Other and unspecified failed attempted abortion, complicated by embolism
O07.8	Other and unspecified failed attempted abortion, with other and unspecified complications
O07.9	Other and unspecified failed attempted abortion, without complication

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## APPENDIX 1: (Continued)

Code	Description
<i>Stillbirth diagnosis codes (ICD-10-AM)</i>	
Z37.1	Outcome of delivery - Single stillbirth
Z37.3	Outcome of delivery - Twins, 1 liveborn and 1 stillborn
Z37.4	Outcome of delivery - Twins, both stillborn
Z37.6	Outcome of delivery - Other multiple births, some liveborn
Z37.7	Outcome of delivery - Other multiple births, all stillborn
Z37.9	Outcome of delivery - Unspecified
<i>Livebirth diagnosis codes (ICD-10-AM)</i>	
Z37.0	Outcome of delivery - Single livebirth
Z37.2	Outcome of delivery - Twins, both liveborn
Z37.5	Outcome of delivery - Other multiple births, all liveborn
<i>Congenital condition diagnosis codes (ICD-10-AM)</i>	
O35.0	Maternal care for known or suspected fetal abnormality - CNS
O35.1	Maternal care for known or suspected fetal abnormality - chromosomal
O35.2	Maternal care for known or suspected fetal abnormality - hereditary
O36.2	Maternal care for other known or suspected fetal problems - hydrops fetalis

Source: NSW Register of Congenital Conditions (RoCC) 2009 and NSW Admitted Patient Data Collection (APD) 2009–2010, Centre for Epidemiology and Evidence, NSW Ministry of Health.

## Appendix 2: ACHI and ICD-10-AM codes for baby inclusion criteria

Code	Description
<i>UDT and talipes repair procedure codes (ACHI)</i>	
37803-00	Repair of undescended testes
37803-01	Repair of undescended testes
49718-01	Repair of talipes
49724-00	Repair of talipes
49724-01	Repair of talipes
49727-00	Repair of talipes
50321-00	Repair of talipes
50324-00	Repair of talipes
50324-01	Repair of talipes
50327-00	Repair of talipes
<i>Congenital condition diagnosis codes (ICD-10-AM)</i>	
D18.1	Lymphangioma, any site
D56.0	Alpha thalassaemia
D56.1	Beta thalassaemia
D82.1	Di George's syndrome
E03.0	Congenital hypothyroidism with diffuse goitre
E03.1	Congenital hypothyroidism without goitre
E70.0	Classical phenylketonuria
E70.1	Other hyperphenylalaninaemias
E84	Cystic fibrosis
E84.0	Cystic fibrosis
E84.1	Cystic fibrosis
E84.8	Cystic fibrosis
E84.9	Cystic fibrosis
P83.2	Hydrops fetalis not due to haemolytic disease

Source: NSW Register of Congenital Conditions (RoCC) 2009 and NSW Admitted Patient Data Collection (APD) 2009–2010, Centre for Epidemiology and Evidence, NSW Ministry of Health.

## Appendix 3: Non-contributory and minor congenital conditions

ICD10-AM Code	Description	Group
Q53.1	Undescended testicle, unilateral	Major only with surgery
Q53.10	Undescended testicle, unilateral	Major only with surgery
Q53.11	Undescended testicle, unilateral	Major only with surgery
Q53.12	Undescended testicle, unilateral	Major only with surgery
Q53.13	Undescended testicle, unilateral	Major only with surgery
Q53.2	Undescended testicle, bilateral	Major only with surgery
Q53.20	Undescended testicle, bilateral	Major only with surgery
Q53.21	Undescended testicle, bilateral	Major only with surgery
Q53.22	Undescended testicle, bilateral	Major only with surgery
Q53.23	Undescended testicle, bilateral	Major only with surgery
Q53.9	Undescended testicle, unspecified laterality	Major only with surgery
Q53.90	Undescended testicle, unspecified laterality	Major only with surgery
Q53.91	Undescended testicle, unspecified laterality	Major only with surgery
Q53.92	Undescended testicle, unspecified laterality	Major only with surgery
Q53.93	Undescended testicle, unspecified laterality	Major only with surgery
Q66.0	Talipes equinovarus	Major only with surgery
Q66.00	Talipes equinovarus, unspecified	Major only with surgery
Q66.01	Structural talipes equinovarus	Major only with surgery
Q66.1	Talipes calcaneovarus	Major only with surgery
Q66.4	Talipes calcanovalgus	Major only with surgery
Q17.2	Microtia	Minor
Q26.1	Persistent left superior vena cava	Minor
Q27.0	Congenital absence and hypoplasia of umbilical artery	Minor
Q27.01	Congenital absence of umbilical artery	Minor
Q27.02	Congenital hypoplasia of umbilical artery	Minor
Q35.7	Cleft uvula	Minor
Q38.2	Macroglossia	Minor
Q52.4	Other congenital malformations of vagina	Minor
Q54.4	Congenital chordee	Minor
Q55.6	Other congenital malformations of penis	Minor
Q55.60	Congenital malformation of penis, unspecified	Minor
Q55.61	Congenital circumcision	Minor
Q55.63	Congenital curvature of penis	Minor
Q55.64	Congenital hooded prepuce	Minor
Q55.69	Other congenital malformations of penis	Minor
Q63.2	Ectopic kidney	Minor
Q63.20	Ectopic kidney	Minor
Q63.21	Ectopic kidney	Minor
Q63.22	Ectopic kidney	Minor
Q63.23	Ectopic kidney	Minor
Q63.29	Ectopic kidney	Minor
Q64.41	Cyst of urachus	Minor
Q64.42	Patent urachus	Minor
Q66.5	Congenital pes planus	Minor
Q66.7	Pes cavus	Minor
Q70.1	Webbed fingers	Minor
Q70.3	Webbed toes	Minor
Q74.81	Brachydactyly	Minor
Q76.5	Cervical rib	Minor
Q76.63	Accessory rib	Minor
Q95	Balanced rearrangements and structural markers, not elsewhere classified	Minor
Q04.61	Single congenital cerebral cyst	Non-contributory
Q04.62	Multiple congenital cerebral cysts	Non-contributory
Q10.0	Congenital ptosis	Non-contributory
Q10.1	Congenital ectropion	Non-contributory
Q10.2	Congenital entropion	Non-contributory
Q10.5	Congenital stenosis and stricture of lacrimal duct	Non-contributory
Q10.6	Other congenital malformations of lacrimal apparatus	Non-contributory
Q17.0	Accessory auricle	Non-contributory
Q17.01	Accessory auricle	Non-contributory

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APPENDIX 3: (Continued)

ICD10-AM Code	Description	Group
Q17.02	Ear tag	Non-contributory
Q17.1	Macrotia	Non-contributory
Q17.3	Other misshapen ear	Non-contributory
Q17.4	Misplaced ear	Non-contributory
Q17.5	Prominent ear	Non-contributory
Q18.0	Sinus, fistula and cyst of branchial cleft	Non-contributory
Q18.1	Preauricular sinus and cyst	Non-contributory
Q18.2	Other brachial cleft malformations	Non-contributory
Q18.3	Webbing of neck	Non-contributory
Q18.4	Macrostomia	Non-contributory
Q18.5	Microstomia	Non-contributory
Q18.6	Macrocheilia	Non-contributory
Q18.7	Microcheilia	Non-contributory
Q18.8	Other specified congenital malformations of face and neck	Non-contributory
Q18.9	Congenital malformation of face and neck, unspecified	Non-contributory
Q22.2	Congenital pulmonary valve insufficiency	Non-contributory
Q23.1	Congenital insufficiency of aortic valve	Non-contributory
Q23.3	Congenital mitral insufficiency	Non-contributory
Q24.6	Congenital heart block	Non-contributory
Q24.8	Other specified congenital malformations of heart	Non-contributory
Q24.82	Congenital cardiomegaly	Non-contributory
Q24.84	Congenital diverticulum of heart	Non-contributory
Q24.85	Congenital malformation of pericardium, not elsewhere classified	Non-contributory
Q27.4	Congenital phlebectasia	Non-contributory
Q28.8	Other specified congenital malformations of circulatory system	Non-contributory
Q30.3	Congenital perforated nasal septum	Non-contributory
Q31.3	Laryngocele	Non-contributory
Q31.5	Congenital laryngomalacia	Non-contributory
Q32.0	Congenital tracheomalacia	Non-contributory
Q32.2	Congenital bronchomalacia	Non-contributory
Q33.0	Congenital cystic lung	Non-contributory
Q33.00	Congenital cystic lung, unspecified	Non-contributory
Q33.02	Congenital honeycomb lung	Non-contributory
Q33.03	Congenital single cyst of lung	Non-contributory
Q33.04	Congenital polycystic lung	Non-contributory
Q33.05	Congenital pulmonary lymphangiectasis	Non-contributory
Q33.09	Other congenital cystic lung	Non-contributory
Q33.4	Congenital bronchiectasis	Non-contributory
Q34.1	Congenital cyst of mediastinum	Non-contributory
Q38.1	Ankyloglossia	Non-contributory
Q38.7	Pharyngeal pouch	Non-contributory
Q39.5	Congenital dilatation of oesophagus	Non-contributory
Q39.6	Diverticulum of oesophagus	Non-contributory
Q39.82	Oesophageal dysmotility	Non-contributory
Q39.84	Congenital displacement of oesophagus	Non-contributory
Q39.85	Congenital duplication cyst of oesophagus	Non-contributory
Q40.0	Congenital hypertrophic pyloric stenosis	Non-contributory
Q40.1	Congenital hiatus hernia	Non-contributory
Q43.0	Meckel's diverticulum	Non-contributory
Q43.2	Other congenital functional disorders of colon	Non-contributory
Q43.8	Other specified congenital malformations of intestine	Non-contributory
Q43.82	Congenital transposition of intestine	Non-contributory
Q43.89	Other specified congenital malformations of intestine	Non-contributory
Q43.9	Congenital malformation of intestine, unspecified	Non-contributory
Q44.4	Choledochal cyst	Non-contributory
Q44.6	Cystic disease of liver	Non-contributory
Q45.2	Congenital pancreatic cyst	Non-contributory
Q45.84	Congenital mesenteric cyst, not elsewhere classified	Non-contributory
Q45.85	Congenital ectopic digestive organs, not elsewhere classified	Non-contributory

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APPENDIX 3: (Continued)

ICD10-AM Code	Description	Group
Q45.89	Other specified congenital malformations of digestive system	Non-contributory
Q45.9	Congenital malformation of digestive system, unspecified	Non-contributory
Q50.1	Developmental ovarian cyst	Non-contributory
Q50.10	Developmental ovarian cyst	Non-contributory
Q50.11	Developmental ovarian cyst	Non-contributory
Q50.12	Developmental ovarian cyst	Non-contributory
Q50.2	Congenital torsion of ovary	Non-contributory
Q50.30	Congenital malformation of ovary, unspecified	Non-contributory
Q50.31	Ovarian streak	Non-contributory
Q50.4	Embryonic cyst of fallopian tube	Non-contributory
Q50.5	Embryonic cyst of broad ligament	Non-contributory
Q51.6	Embryonic cyst of cervix	Non-contributory
Q51.9	Congenital malformation of uterus and cervix, unspecified	Non-contributory
Q52.3	Imperforate hymen	Non-contributory
Q52.40	Congenital malformation of vagina, unspecified	Non-contributory
Q52.42	Embryonic cyst of vagina	Non-contributory
Q52.43	Congenital cyst of canal of Nuck	Non-contributory
Q52.49	Other congenital malformations of vagina	Non-contributory
Q52.5	Fusion of labia	Non-contributory
Q55.20	Congenital malformation of testis and scrotum, unspecified	Non-contributory
Q55.21	Retractile testis	Non-contributory
Q55.8	Other specified congenital malformations of male genital organs	Non-contributory
Q55.9	Congenital malformation of male genital organ, unspecified	Non-contributory
Q61.0	Congenital single renal cyst	Non-contributory
Q61.5	Medullary cystic kidney	Non-contributory
Q61.50	Medullary cystic kidney	Non-contributory
Q61.51	Medullary cystic kidney	Non-contributory
Q61.52	Medullary cystic kidney	Non-contributory
Q62.33	Congenital polyp of ureter	Non-contributory
Q62.7	Congenital vesico-uretero-renal reflux	Non-contributory
Q62.70	Congenital vesico-uretero-renal reflux	Non-contributory
Q62.71	Congenital vesico-uretero-renal reflux	Non-contributory
Q62.72	Congenital vesico-uretero-renal reflux	Non-contributory
Q63.3	Hyperplastic and giant kidney	Non-contributory
Q63.81	Congenital calyceal diverticulum	Non-contributory
Q63.82	Congenital renal calculi	Non-contributory
Q63.89	Other specified congenital malformations of kidney	Non-contributory
Q64.43	Urachal diverticulum	Non-contributory
Q64.49	Other specified malformation of urachus	Non-contributory
Q64.6	Congenital diverticulum of bladder	Non-contributory
Q64.71	Congenital anterior urethral diverticulum	Non-contributory
Q64.72	Congenital prolapse of bladder (mucosa)	Non-contributory
Q64.77	Megacystitis-megaureter syndrome	Non-contributory
Q64.78	Congenital urethral syringocele	Non-contributory
Q64.8	Other specified congenital malformations of urinary system	Non-contributory
Q64.9	Congenital malformation of urinary system, unspecified	Non-contributory
Q65.8	Other congenital deformities of hip	Non-contributory
Q65.81	Congenital coxa valga	Non-contributory
Q65.82	Congenital coxa vara	Non-contributory
Q65.89	Other congenital deformities of the hip	Non-contributory
Q65.9	Congenital deformity of hip, unspecified	Non-contributory
Q66.02	Positional talipes equinovarus	Non-contributory
Q66.2	Metatarsus varus	Non-contributory
Q66.3	Other congenital varus deformities of feet	Non-contributory
Q66.6	Other congenital valgus deformities of feet	Non-contributory
Q66.82	Congenital hammer toe	Non-contributory
Q66.83	Congenital shortening of Achilles tendon	Non-contributory
Q66.84	Congenital spade-like foot	Non-contributory
Q66.89	Other congenital deformities of feet	Non-contributory

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APPENDIX 3: (Continued)

ICD10-AM Code	Description	Group
Q66.9	Congenital deformity of feet, unspecified	Non-contributory
Q67.0	Facial asymmetry	Non-contributory
Q67.1	Compression of facies	Non-contributory
Q67.2	Dolichocephaly	Non-contributory
Q67.3	Plagiocephaly	Non-contributory
Q67.41	Depressions in skull	Non-contributory
Q67.42	Deviation of nasal septum, congenital	Non-contributory
Q67.43	Congenital hemifacial atrophy	Non-contributory
Q67.44	Congenital hemifacial hypertrophy	Non-contributory
Q68.0	Congenital deformities of sternocleidomastoid muscle	Non-contributory
Q68.00	Congenital deformity of sternocleidomastoid muscle, unspecified	Non-contributory
Q68.01	Congenital torticollis	Non-contributory
Q68.02	Congenital contracture of sternocleidomastoid muscle	Non-contributory
Q68.03	Congenital sternomastoid lesion	Non-contributory
Q68.2	Congenital deformity of knee	Non-contributory
Q68.3	Congenital bowing of femur	Non-contributory
Q68.4	Congenital bowing of tibia and fibula	Non-contributory
Q68.5	Congenital bowing of long bones of leg, unspecified	Non-contributory
Q68.8	Other specified congenital musculoskeletal deformities	Non-contributory
Q74.03	Triphalangeal thumb	Non-contributory
Q74.05	Humeroulnar synostosis	Non-contributory
Q74.06	Humeroradial synostosis	Non-contributory
Q74.08	Sprengel's deformity	Non-contributory
Q74.1	Congenital malformation of knee	Non-contributory
Q74.10	Congenital malformation of knee, unspecified	Non-contributory
Q74.11	Congenital absence of patella	Non-contributory
Q74.12	Congenital dislocation of patella	Non-contributory
Q74.13	Rudimentary patella	Non-contributory
Q74.14	Genu valgum	Non-contributory
Q74.15	Genu varum	Non-contributory
Q74.19	Other congenital malformations of knee	Non-contributory
Q74.2	Other congenital malformations of lower limb(s), including pelvic girdle	Non-contributory
Q74.20	Congenital malformation of lower limb(s), including pelvic girdle	Non-contributory
Q74.21	Other congenital malformations of pelvis, not elsewhere classified	Non-contributory
Q74.22	Other congenital malformations of thigh, not elsewhere classified	Non-contributory
Q74.23	Other congenital malformations of lower leg, not elsewhere classified	Non-contributory
Q74.24	Other congenital malformations of ankle, not elsewhere classified	Non-contributory
Q74.25	Other congenital malformations of foot, not elsewhere classified	Non-contributory
Q74.26	Other congenital malformations of toe(s), not elsewhere classified	Non-contributory
Q74.82	Congenital overgrowth of limbs	Non-contributory
Q74.83	Congenital undergrowth of limbs	Non-contributory
Q74.84	Congenital limb asymmetry	Non-contributory
Q74.89	Other specified congenital malformations of limb(s)	Non-contributory
Q74.9	Unspecified congenital malformation of limb(s)	Non-contributory
Q75.2	Hypertelorism	Non-contributory
Q75.3	Macrocephaly	Non-contributory
Q75.31	Macrocephaly	Non-contributory
Q75.39	Macrocephaly	Non-contributory
Q76.0	Spina bifida occulta	Non-contributory
Q76.44	Congenital lordosis, postural	Non-contributory
Q76.49	Other congenital malformation of spine	Non-contributory
Q78.6	Multiple congenital exostoses	Non-contributory
Q79.50	Congenital malformation of abdominal wall, unspecified	Non-contributory
Q79.52	Diastasis recti	Non-contributory
Q79.59	Other congenital malformations of abdominal wall	Non-contributory
Q82.2	Mastocytosis	Non-contributory
Q82.4	Ectodermal dysplasia (anhidrotic)	Non-contributory
Q82.5	Congenital non-neoplastic naevus	Non-contributory if < 4 cm
Q82.8	Other specified congenital malformations of skin	Non-contributory

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APPENDIX 3: (Continued)

ICD10-AM Code	Description	Group
Q82.9	Congenital malformation of skin, unspecified	Non-contributory
Q83.3	Accessory nipple	Non-contributory
Q84.1	Congenital morphological disturbances of hair, not elsewhere classified	Non-contributory
Q84.2	Other congenital malformations of hair	Non-contributory
Q84.4	Congenital leukonychia	Non-contributory
Q84.5	Enlarged and hypertrophic nails	Non-contributory
Q84.6	Other congenital malformations of nails	Non-contributory
Q84.60	Congenital malformations of nails, unspecified	Non-contributory
Q84.61	Congenital clubnail	Non-contributory
Q84.62	Congenital koilonychias	Non-contributory
Q84.69	Other congenital malformations of nails	Non-contributory
Q84.89	Other specified congenital malformations of integument	Non-contributory
Q84.9	Congenital malformation of integument, unspecified	Non-contributory
Q85.0	Neurofibromatosis (nonmalignant)	Non-contributory
Q86.0	Fetal alcohol syndrome (dysmorphic)	Non-contributory
Q86.1	Fetal hydantoin syndrome	Non-contributory
Q86.2	Dysmorphism due to warfarin	Non-contributory
Q86.8	Other congenital malformation syndromes due to known exogenous causes	Non-contributory
Q86.81	Other congenital malformation syndromes due to known exogenous causes	Non-contributory
Q86.82	Other congenital malformation syndromes due to known exogenous causes	Non-contributory
Q86.83	Other congenital malformation syndromes due to known exogenous causes	Non-contributory
Q86.84	Other congenital malformation syndromes due to known exogenous causes	Non-contributory
Q86.85	Other congenital malformation syndromes due to known exogenous causes	Non-contributory
Q86.86	Other congenital malformation syndromes due to known exogenous causes	Non-contributory
Q86.89	Other congenital malformation syndromes due to known exogenous causes	Non-contributory
Q89.23	Persistent thyroglossal cyst	Non-contributory
Q89.24	Thyroglossal cyst	Non-contributory
Q89.71	Dysmorphic features	Non-contributory
Q95.0	Balanced rearrangements and structural markers, not elsewhere classified	Non-contributory
Q95.1	Balanced rearrangements and structural markers, not elsewhere classified	Non-contributory
Q95.2	Balanced rearrangements and structural markers, not elsewhere classified	Non-contributory
Q95.3	Balanced rearrangements and structural markers, not elsewhere classified	Non-contributory
Q95.4	Balanced rearrangements and structural markers, not elsewhere classified	Non-contributory
Q95.5	Balanced rearrangements and structural markers, not elsewhere classified	Non-contributory
Q95.8	Balanced rearrangements and structural markers, not elsewhere classified	Non-contributory
Q95.9	Balanced rearrangements and structural markers, not elsewhere classified	Non-contributory
Q25.0	Patent ductus arteriosus	Non-contributory or minor
Q33.6	Hypoplasia and dysplasia of lung	Non-contributory or minor

Source: NSW Register of Congenital Conditions (RoCC) 2009 and NSW Admitted Patient Data Collection (APD) 2009–2010, Centre for Epidemiology and Evidence, NSW Ministry of Health.

## Appendix 4: BPA and ICD-10-AM Code Groups and Categories for Congenital Conditions

Code	Type	Description	Specific condition group	Major diagnostic category
759210	BPA	Absence of aplasia of thyroid gland	Absence or aplasia of thyroid gland	Other and unspecified anomalies
74400	BPA	Absence or stricture of auditory canal	Absence-stricture auditory canal	Ear, face and neck
74843	BPA	Adenomatoid malformation of lung	Adenomatoid malformation of lung	Respiratory system
75142	BPA	Adhesions or bands of omentum & peritoneum	Adhesions or bands of omentum & peritoneum	Gastrointestinal system
74224	BPA	Agyria & lissencephaly	Agyria and microgyria	Nervous system
74225	BPA	Microgyria	Agyria and microgyria	Nervous system
74001	BPA	Acrania	Anencephaly	Nervous system
74002	BPA	Anencephaly	Anencephaly	Nervous system
74353	BPA	Anomaly of choroid - specified NEC	Anomalies of choroid	Eye
74352	BPA	Anomaly of optic disc - specified NEC	Anomalies of optic disc	Eye
74351	BPA	Anomaly of retina - specified NEC	Anomalies of retina	Eye
75630	BPA	Absence of ribs	Anomalies of ribs and sternum	Musculoskeletal system
75632	BPA	Fused ribs	Anomalies of ribs and sternum	Musculoskeletal system
75634	BPA	Anomaly of ribs - other specified NEC	Anomalies of ribs and sternum	Musculoskeletal system
75637	BPA	Anomaly of chest wall or thoracic cage - other specified NEC	Anomalies of ribs and sternum	Musculoskeletal system
75638	BPA	Anomaly of sternum - other specified NEC	Anomalies of ribs and sternum	Musculoskeletal system
75612	BPA	Kyphosis	Anomalies of spinal vertebrae	Musculoskeletal system
756141	BPA	Hemivertebrae - cervical	Anomalies of spinal vertebrae	Musculoskeletal system
756148	BPA	Anomaly of vertebrae - cervical - other specified NEC	Anomalies of spinal vertebrae	Musculoskeletal system
756151	BPA	Hemivertebrae - thoracic	Anomalies of spinal vertebrae	Musculoskeletal system
756158	BPA	Anomaly of vertebrae - thoracic - other specified NEC	Anomalies of spinal vertebrae	Musculoskeletal system
756161	BPA	Hemivertebrae - lumbar	Anomalies of spinal vertebrae	Musculoskeletal system
756168	BPA	Anomaly of vertebrae - lumbar - other specified NEC	Anomalies of spinal vertebrae	Musculoskeletal system
756170	BPA	Absence of vertebrae - sacrum &/or coccyx	Anomalies of spinal vertebrae	Musculoskeletal system
756172	BPA	Dysplasia of sacrum &/or coccyx	Anomalies of spinal vertebrae	Musculoskeletal system
756178	BPA	Anomaly of vertebrae - sacrococcygeal - other specified NEC	Anomalies of spinal vertebrae	Musculoskeletal system
75618	BPA	Anomaly of vertebrae - other specified NEC	Anomalies of spinal vertebrae	Musculoskeletal system
74350	BPA	Anomaly of vitreous humour - specified NEC	Anomalies of vitreous humour	Eye
74300	BPA	Anophthalmos/agenesis of eye	Anophthalmos-microphthalmos	Eye
74301	BPA	Cryptophthalmos	Anophthalmos-microphthalmos	Eye
74310	BPA	Microphthalmos - NOS	Anophthalmos-microphthalmos	Eye
75280	BPA	Absence of testicle	Aplasia or hypoplasia of testes or scrotum	Genitourinary system
752810	BPA	Aplasia of testicle	Aplasia or hypoplasia of testes or scrotum	Genitourinary system
752813	BPA	Hypoplasia of testicle	Aplasia or hypoplasia of testes or scrotum	Genitourinary system
752814	BPA	Hypoplasia of scrotum	Aplasia or hypoplasia of testes or scrotum	Genitourinary system
75580	BPA	Arthrogryposis multiplex congenita	Arthrogryposis	Musculoskeletal system
75582	BPA	Arthrogryposis - other specified NEC & NOS	Arthrogryposis	Musculoskeletal system
759001	BPA	Ivemark's syndrome - bilateral right sided sequence	Asplenia	Other and unspecified anomalies
74630	BPA	Atresia of aortic valve	Atresia of aortic valve	Cardiovascular system
74642	BPA	Hypoplasia or dysplasia of aortic valve	Atresia of aortic valve	Cardiovascular system
751651	BPA	Atresia of hepatic or bile ducts	Atresia of hepatic or bile ducts	Gastrointestinal system
747300	BPA	Absence or agenesis of pulmonary artery	Atresia of pulmonary artery	Cardiovascular system
747301	BPA	Atresia of pulmonary artery	Atresia of pulmonary artery	Cardiovascular system
74731	BPA	Atresia of pulmonary artery with septal defect	Atresia of pulmonary artery	Cardiovascular system
74735	BPA	Hypoplasia of pulmonary artery	Atresia of pulmonary artery	Cardiovascular system
74600	BPA	Atresia of pulmonary valve	Atresia of pulmonary valve	Cardiovascular system
74604	BPA	Absence of pulmonary valve	Atresia of pulmonary valve	Cardiovascular system
74610	BPA	Atresia of tricuspid valve	Atresia of tricuspid valve	Cardiovascular system
74613	BPA	Hypoplasia or dysplasia of tricuspid valve	Atresia of tricuspid valve	Cardiovascular system
751230	BPA	Absence of anus - with fistula	Atresia-stenosis of anus (includes with fistula)	Gastrointestinal system
751231	BPA	Atresia of anus - with fistula	Atresia-stenosis of anus (includes with fistula)	Gastrointestinal system
751240	BPA	Absence of anus - no mention of fistula	Atresia-stenosis of anus (includes with fistula)	Gastrointestinal system
751241	BPA	Atresia of anus - no mention of fistula	Atresia-stenosis of anus (includes with fistula)	Gastrointestinal system
751242	BPA	Stenosis of anus - no mention of fistula	Atresia-stenosis of anus (includes with fistula)	Gastrointestinal system
751201	BPA	Atresia of large intestine	Atresia-stenosis of large intestine (includes with fistula)	Gastrointestinal system
751221	BPA	Atresia of rectum - no mention of fistula	Atresia-stenosis of large intestine (includes with fistula)	Gastrointestinal system
751101	BPA	Atresia of duodenum	Atresia-stenosis of small intestine (includes with fistula)	Gastrointestinal system
751102	BPA	Stenosis of duodenum	Atresia-stenosis of small intestine (includes with fistula)	Gastrointestinal system

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APPENDIX 4: (Continued)

Code	Type	Description	Specific condition group	Major diagnostic category
751111	BPA	Atresia of jejunum	Atresia-stenosis of small intestine (includes with fistula)	Gastrointestinal system
751121	BPA	Atresia of ileum	Atresia-stenosis of small intestine (includes with fistula)	Gastrointestinal system
74550	BPA	Patent foramen ovale	Atrial septal defect	Cardiovascular system
74551	BPA	Atrial septal defect - ostium secundum defect	Atrial septal defect	Cardiovascular system
74558	BPA	Atrial septal defect - other specified NEC	Atrial septal defect	Cardiovascular system
74559	BPA	Atrial septal defect - NOS	Atrial septal defect	Cardiovascular system
74560	BPA	Endocardial cushion defect - ostium primum defect	Atrial septal defect	Cardiovascular system
74561	BPA	Common atrium	Atrial septal defect	Cardiovascular system
74562	BPA	Common A-V canal type ventricular septal defect - complete	Atrioventricular septal defect	Cardiovascular system
74563	BPA	Common A-V canal - incomplete	Atrioventricular septal defect	Cardiovascular system
75831	BPA	Cri du chat syndrome	Autosomal chromosome deletions	Chromosomal anomalies
75832	BPA	Wolff Hirschorn syndrome	Autosomal chromosome deletions	Chromosomal anomalies
75833	BPA	Deletion of long arm of 13	Autosomal chromosome deletions	Chromosomal anomalies
75834	BPA	Deletion of long arm of 17 or 18	Autosomal chromosome deletions	Chromosomal anomalies
75835	BPA	Deletion of short arm of 17 or 18	Autosomal chromosome deletions	Chromosomal anomalies
75838	BPA	Loss of autosomal material - other specified NEC	Autosomal chromosome deletions	Chromosomal anomalies
74641	BPA	Bicuspid aortic valve	Bicuspid aortic valve	Cardiovascular system
74320	BPA	Buphthalmos	Buphthalmos-congenital glaucoma	Eye
759893	BPA	CHARGE association	CHARGE syndrome	Congenital malformation syndromes
74800	BPA	Choanal atresia - atresia of nares	Choanal atresia and stenosis	Respiratory system
74801	BPA	Stenosis of nares	Choanal atresia and stenosis	Respiratory system
75641	BPA	Chondrodysplasia	Chondrodystrophy	Musculoskeletal system
75643	BPA	Achondroplastic dwarfism	Chondrodystrophy	Musculoskeletal system
756442	BPA	Thanatophoric dysplasia/dwarfism	Chondrodystrophy	Musculoskeletal system
756448	BPA	Dwarfing syndromes - other specified NEC	Chondrodystrophy	Musculoskeletal system
75646	BPA	Spondyloepiphyseal dysplasia	Chondrodystrophy	Musculoskeletal system
749200	BPA	Cleft lip & palate - unilateral cleft lip - cleft hard palate	Cleft lip and palate	Gastrointestinal system
749201	BPA	Cleft lip & palate - unilateral cleft lip - cleft soft palate	Cleft lip and palate	Gastrointestinal system
749202	BPA	Cleft lip & palate - unilateral cleft lip - cleft hard & soft palate	Cleft lip and palate	Gastrointestinal system
749209	BPA	Cleft lip & palate - unilateral cleft lip - cleft palate NOS	Cleft lip and palate	Gastrointestinal system
749212	BPA	Cleft lip & palate - bilateral cleft lip - cleft hard & soft palate	Cleft lip and palate	Gastrointestinal system
749219	BPA	Cleft lip & palate - bilateral cleft lip - cleft palate NOS	Cleft lip and palate	Gastrointestinal system
749221	BPA	Cleft lip & palate - central cleft lip - cleft soft palate	Cleft lip and palate	Gastrointestinal system
749222	BPA	Cleft lip & palate - central cleft lip - cleft hard & soft palate	Cleft lip and palate	Gastrointestinal system
749229	BPA	Cleft lip & palate - central cleft lip - cleft palate NOS	Cleft lip and palate	Gastrointestinal system
749292	BPA	Cleft lip & palate - cleft lip NOS - cleft hard & soft palate	Cleft lip and palate	Gastrointestinal system
749299	BPA	Cleft lip & palate - cleft lip NOS - cleft palate NOS	Cleft lip and palate	Gastrointestinal system
74910	BPA	Cleft lip - unilateral	Cleft lip only	Gastrointestinal system
74911	BPA	Cleft lip - bilateral	Cleft lip only	Gastrointestinal system
74919	BPA	Cleft lip - NOS	Cleft lip only	Gastrointestinal system
74901	BPA	Cleft palate - hard - bilateral	Cleft palate only	Gastrointestinal system
74902	BPA	Cleft palate - hard - central	Cleft palate only	Gastrointestinal system
74905	BPA	Cleft palate - soft - bilateral	Cleft palate only	Gastrointestinal system
74906	BPA	Cleft palate - soft - central	Cleft palate only	Gastrointestinal system
74907	BPA	Cleft palate - soft - NOS	Cleft palate only	Gastrointestinal system
749081	BPA	Cleft palate - submucous	Cleft palate only	Gastrointestinal system
749082	BPA	Cleft gum	Cleft palate only	Gastrointestinal system
74909	BPA	Cleft palate - NOS	Cleft palate only	Gastrointestinal system
74931	BPA	Cleft palate - hard & soft palate - bilateral	Cleft palate only	Gastrointestinal system
74932	BPA	Cleft palate - hard & soft palate - central	Cleft palate only	Gastrointestinal system
74939	BPA	Cleft palate - hard & soft palate - NOS	Cleft palate only	Gastrointestinal system
74710	BPA	Coarctation of aorta - preductal	Coarctation of aorta	Cardiovascular system
74711	BPA	Coarctation of aorta - postductal	Coarctation of aorta	Cardiovascular system
74712	BPA	Interrupted aortic arch	Coarctation of aorta	Cardiovascular system
74713	BPA	Coarctation of aorta - juxtaductal	Coarctation of aorta	Cardiovascular system
74719	BPA	Coarctation of aorta - NOS	Coarctation of aorta	Cardiovascular system
74343	BPA	Coloboma of iris	Coloboma of iris	Eye
74500	BPA	Truncus arteriosus	Common arterial trunk	Cardiovascular system
74501	BPA	Aortic septal defect - aortopulmonary window	Common arterial trunk	Cardiovascular system
74783	BPA	Absent ductus arteriosus	Common arterial trunk	Cardiovascular system
7453	BPA	Common ventricle	Common ventricle	Cardiovascular system

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APPENDIX 4: (Continued)

Code	Type	Description	Specific condition group	Major diagnostic category
74332	BPA	Congenital cataract	Congenital cataract	Eye
75430	BPA	Dislocation of hip	Congenital dislocation of hips	Musculoskeletal system
742300	BPA	Congenital hydrocephalus - stenosis or obstruction aqueduct of Sylvius	Congenital hydrocephalus	Nervous system
742309	BPA	Congenital hydrocephalus - anomaly of aqueduct of Sylvius - NOS	Congenital hydrocephalus	Nervous system
74231	BPA	Congenital hydrocephalus - atresia of foramina of Magendie & Luchka - Dandy Walker syndrome	Congenital hydrocephalus	Nervous system
74232	BPA	Congenital hydrocephalus - hydranencephaly	Congenital hydrocephalus	Nervous system
74233	BPA	Congenital hydrocephalus - communicating NOS	Congenital hydrocephalus	Nervous system
74234	BPA	Congenital hydrocephalus - associated with other brain deformity	Congenital hydrocephalus	Nervous system
74239	BPA	Congenital hydrocephalus - NOS	Congenital hydrocephalus	Nervous system
24399	BPA	Congenital hypothyroidism	Congenital hypothyroidism	Other and unspecified anomalies
756000	BPA	Craniosynostosis - coronal	Craniosynostosis	Musculoskeletal system
756002	BPA	Craniosynostosis - sagittal	Craniosynostosis	Musculoskeletal system
756003	BPA	Craniosynostosis - metopic	Craniosynostosis	Musculoskeletal system
756005	BPA	Trigonocephaly	Craniosynostosis	Musculoskeletal system
756009	BPA	Craniosynostosis - NOS	Craniosynostosis	Musculoskeletal system
2770	BPA	Cystic fibrosis	Cystic fibrosis	Other and unspecified anomalies
75311	BPA	Polycystic kidneys - infantile type	Cystic kidney disease	Genitourinary system
75312	BPA	Polycystic kidneys - adult type	Cystic kidney disease	Genitourinary system
75313	BPA	Polycystic kidneys - NOS	Cystic kidney disease	Genitourinary system
75316	BPA	Multicystic renal dysplasia	Cystic kidney disease	Genitourinary system
75319	BPA	Cystic kidney disease - NOS	Cystic kidney disease	Genitourinary system
74680	BPA	Dextrocardia without situs inversus	Dextrocardia	Cardiovascular system
75661	BPA	Diaphragmatic hernia	Diaphragmatic hernia	Musculoskeletal system
753310	BPA	Double kidney	Duplex kidney and/or collecting system	Genitourinary system
753312	BPA	Pyelon duplex or triplex	Duplex kidney and/or collecting system	Genitourinary system
75341	BPA	Accessory ureter	Duplex kidney and/or collecting system	Genitourinary system
75342	BPA	Ectopic ureter	Duplex kidney and/or collecting system	Genitourinary system
7462	BPA	Ebstein's anomaly	Ebstein's anomaly	Cardiovascular system
751530	BPA	Ectopic anus - anterior	Ectopic anus	Gastrointestinal system
751539	BPA	Ectopic anus - NOS	Ectopic anus	Gastrointestinal system
759212	BPA	Ectopic thyroid gland	Ectopic thyroid gland	Other and unspecified anomalies
742000	BPA	Encephalocele - no mention of hydrocephalus - occipital	Encephalocele	Nervous system
742001	BPA	Encephalocele - with hydrocephalus - occipital	Encephalocele	Nervous system
742010	BPA	Encephalocele - no mention of hydrocephalus - parietal	Encephalocele	Nervous system
742020	BPA	Encephalocele - no mention of hydrocephalus - frontal	Encephalocele	Nervous system
742090	BPA	Encephalocele - no mention of hydrocephalus - site NOS	Encephalocele	Nervous system
742091	BPA	Encephalocele - with hydrocephalus - site NOS	Encephalocele	Nervous system
75670	BPA	Exomphalos	Exomphalos	Musculoskeletal system
75671	BPA	Gastroschisis	Gastroschisis	Musculoskeletal system
75130	BPA	Total intestinal aganglionosis	Hirschsprungs Disease	Gastrointestinal system
75131	BPA	Hirschsprung's disease - long segment	Hirschsprungs Disease	Gastrointestinal system
75132	BPA	Hirschsprung's disease - short segment	Hirschsprungs Disease	Gastrointestinal system
75133	BPA	Hirschsprung's disease - NOS	Hirschsprungs Disease	Gastrointestinal system
74226	BPA	Holoprosencephaly - arhinencephaly	Holoprosencephaly	Nervous system
753322	BPA	Horseshoe kidney	Horseshoe kidney	Genitourinary system
75320	BPA	Hydronephrosis	Hydronephrosis	Genitourinary system
7333	BPA	Hydrops fetalis due to isoimmunization	Hydrops due to isoimmunization	Other and unspecified anomalies
7780	BPA	Hydrops foetalis not due to isoimmunization	Hydrops not due to isoimmunization	Non-immune hydrops foetalis
753212	BPA	Hydroureter NOS	Hydroureter and megaloureter	Genitourinary system
75322	BPA	Megaloureter - NOS	Hydroureter and megaloureter	Genitourinary system
74382	BPA	Hypoplasia of optic nerves	Hypoplasia of optic nerve	Eye
7467	BPA	Hypoplastic left heart syndrome	Hypoplastic heart	Cardiovascular system
746883	BPA	Hypoplastic left heart	Hypoplastic heart	Cardiovascular system
746884	BPA	Hypoplastic right heart	Hypoplastic heart	Cardiovascular system
746885	BPA	Hypoplastic heart - NOS	Hypoplastic heart	Cardiovascular system
746880	BPA	Hypoplastic left ventricle	Hypoplastic left ventricle	Cardiovascular system
752601	BPA	Hypospadias - 1st degree	Hypospadias	Genitourinary system
752602	BPA	Hypospadias - 2nd degree	Hypospadias	Genitourinary system
752603	BPA	Hypospadias - 3rd degree	Hypospadias	Genitourinary system
752604	BPA	Hypospadias - 4th degree	Hypospadias	Genitourinary system
752609	BPA	Hypospadias - NOS	Hypospadias	Genitourinary system

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APPENDIX 4: (Continued)

Code	Type	Description	Specific condition group	Major diagnostic category
752719	BPA	Pseudohermaphroditism - male - NOS	Indeterminate sex - ambiguous genitalia	Genitourinary system
752781	BPA	Ambiguous genitalia - male	Indeterminate sex - ambiguous genitalia	Genitourinary system
752782	BPA	Ambiguous genitalia - female	Indeterminate sex - ambiguous genitalia	Genitourinary system
75143	BPA	Midgut malrotation	Intestinal malrotation	Gastrointestinal system
75144	BPA	Malrotation - NOS	Intestinal malrotation	Gastrointestinal system
75780	BPA	Cystic hygroma	Lymphangioma	Integumentary system
75781	BPA	Lymphangioma - congenital - NOS	Lymphangioma	Integumentary system
74210	BPA	Microcephalus	Microcephaly	Nervous system
758621	BPA	Noonan syndrome	Noonan syndrome	Congenital malformation syndromes
74818	BPA	Anomaly of nose - other specified NEC	Nose, larynx, trachea, bronchus	Respiratory system
74830	BPA	Anomaly of larynx & supporting cartilage	Nose, larynx, trachea, bronchus	Respiratory system
74833	BPA	Anomaly of trachea - other specified NEC	Nose, larynx, trachea, bronchus	Respiratory system
74834	BPA	Stenosis of bronchus	Nose, larynx, trachea, bronchus	Respiratory system
748358	BPA	Anomaly of bronchus - other specified NEC	Nose, larynx, trachea, bronchus	Respiratory system
750300	BPA	Absence of oesophagus - no mention of fistula	Oesophageal atresia only	Gastrointestinal system
750301	BPA	Atresia of oesophagus - no mention of fistula	Oesophageal atresia only	Gastrointestinal system
750311	BPA	Atresia of oesophagus - with tracheo-oesophageal fistula	Oesophageal atresia with TOF	Gastrointestinal system
75650	BPA	Osteogenesis imperfecta	Osteodystrophy	Musculoskeletal system
75659	BPA	Osteodystrophy - NOS	Osteodystrophy	Musculoskeletal system
75672	BPA	Prune belly syndrome	Other anomalies of abdominal wall	Musculoskeletal system
75678	BPA	Anomaly of abdominal wall - other specified NEC	Other anomalies of abdominal wall	Musculoskeletal system
75910	BPA	Absence of adrenal gland	Other anomalies of adrenal gland	Other and unspecified anomalies
75911	BPA	Hypoplasia of adrenal gland	Other anomalies of adrenal gland	Other and unspecified anomalies
75918	BPA	Anomaly of adrenal gland - other specified NEC	Other anomalies of adrenal gland	Other and unspecified anomalies
74340	BPA	Corneal opacity	Other anomalies of anterior segment	Eye
74345	BPA	Blue sclera	Other anomalies of anterior segment	Eye
74348	BPA	Other anomalies of anterior segment	Other anomalies of anterior segment	Eye
747200	BPA	Absence of aorta	Other anomalies of aorta	Cardiovascular system
747201	BPA	Atresia of aorta	Other anomalies of aorta	Cardiovascular system
74721	BPA	Hypoplasia of aorta	Other anomalies of aorta	Cardiovascular system
74722	BPA	Supra-aortic stenosis	Other anomalies of aorta	Cardiovascular system
74723	BPA	Persistent right aortic arch	Other anomalies of aorta	Cardiovascular system
74725	BPA	Vascular ring of aorta - double aortic arch	Other anomalies of aorta	Cardiovascular system
74726	BPA	Overriding aorta	Other anomalies of aorta	Cardiovascular system
74727	BPA	Aneurysm of aorta	Other anomalies of aorta	Cardiovascular system
74728	BPA	Anomaly of aorta - other specified NEC	Other anomalies of aorta	Cardiovascular system
74632	BPA	Subaortic stenosis	Other anomalies of aortic and mitral valve	Cardiovascular system
74648	BPA	Anomaly of aortic valve - other specified NEC	Other anomalies of aortic and mitral valve	Cardiovascular system
74649	BPA	Anomaly of aortic valve - NOS	Other anomalies of aortic and mitral valve	Cardiovascular system
74650	BPA	Atresia of mitral valve	Other anomalies of aortic and mitral valve	Cardiovascular system
74651	BPA	Stenosis of mitral valve	Other anomalies of aortic and mitral valve	Cardiovascular system
74661	BPA	Hypoplasia or dysplasia of mitral valve	Other anomalies of aortic and mitral valve	Cardiovascular system
74668	BPA	Anomaly of mitral valve - other specified NEC	Other anomalies of aortic and mitral valve	Cardiovascular system
7401	BPA	Craniorachischisis	Other anomalies of brain	Nervous system
74029	BPA	Iniencephaly - unspecified	Other anomalies of brain	Nervous system
742401	BPA	Megalencephaly	Other anomalies of brain	Nervous system
74248	BPA	Anomaly of brain - other specified NEC	Other anomalies of brain	Nervous system
75660	BPA	Absence of diaphragm	Other anomalies of diaphragm	Musculoskeletal system
75662	BPA	Eversion of diaphragm	Other anomalies of diaphragm	Musculoskeletal system
75668	BPA	Anomaly of diaphragm - other specified NEC	Other anomalies of diaphragm	Musculoskeletal system
74401	BPA	Absence of auricle - ear NOS	Other anomalies of ear	Ear, face and neck
74403	BPA	Anomaly of inner ear	Other anomalies of ear	Ear, face and neck
759218	BPA	Anomaly of thyroid gland - other specified NEC	Other anomalies of endocrine glands	Other and unspecified anomalies
759240	BPA	Absence or aplasia of thymus	Other anomalies of endocrine glands	Other and unspecified anomalies
74363	BPA	Anomaly of eyelids - other specified NEC	Other anomalies of eye	Eye
74388	BPA	Other specified anomalies of eye	Other anomalies of eye	Eye
75200	BPA	Absence or agenesis of ovary	Other anomalies of female genitals	Genitourinary system
75201	BPA	Streak ovary	Other anomalies of female genitals	Genitourinary system
75220	BPA	Doubling of uterus - no mention of double cervix & vagina	Other anomalies of female genitals	Genitourinary system
75230	BPA	Absence or agenesis of uterus	Other anomalies of female genitals	Genitourinary system
75234	BPA	Bicornuate uterus	Other anomalies of female genitals	Genitourinary system

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APPENDIX 4: (Continued)

Code	Type	Description	Specific condition group	Major diagnostic category
75238	BPA	Anomaly of uterus - other specified NEC	Other anomalies of female genitals	Genitourinary system
752411	BPA	Atresia of vagina	Other anomalies of female genitals	Genitourinary system
751630	BPA	Agensis of gallbladder	Other anomalies of gallbladder, bile duct, pancreas and liver	Gastrointestinal system
75164	BPA	Anomaly of gallbladder - other specified NEC	Other anomalies of gallbladder, bile duct, pancreas and liver	Gastrointestinal system
751652	BPA	Alagille syndrome - arteriohepatic dysplasia	Other anomalies of gallbladder, bile duct, pancreas and liver	Gastrointestinal system
75172	BPA	Annular pancreas	Other anomalies of gallbladder, bile duct, pancreas and liver	Gastrointestinal system
75173	BPA	Ectopic pancreas	Other anomalies of gallbladder, bile duct, pancreas and liver	Gastrointestinal system
75178	BPA	Anomaly of pancreas - other specified NEC	Other anomalies of gallbladder, bile duct, pancreas and liver	Gastrointestinal system
74748	BPA	Anomaly of great veins - other specified NEC	Other anomalies of great veins	Cardiovascular system
74681	BPA	Levocardia	Other anomalies of heart	Cardiovascular system
74682	BPA	Cor triatriatum	Other anomalies of heart	Cardiovascular system
74683	BPA	Pulmonary infundibular stenosis	Other anomalies of heart	Cardiovascular system
746882	BPA	Ectopic heart	Other anomalies of heart	Cardiovascular system
746886	BPA	Anomaly of coronary artery	Other anomalies of heart	Cardiovascular system
74693	BPA	Congenital heart disease - cyanotic	Other anomalies of heart	Cardiovascular system
74699	BPA	Anomaly of heart - NOS	Other anomalies of heart	Cardiovascular system
75148	BPA	Anomaly of intestinal fixation - other specified NEC	Other anomalies of intestinal fixation	Gastrointestinal system
753321	BPA	Fused kidney	Other anomalies of kidney and collecting system	Genitourinary system
75340	BPA	Absence of ureter	Other anomalies of kidney and collecting system	Genitourinary system
75348	BPA	Anomaly of ureter - other specified NEC	Other anomalies of kidney and collecting system	Genitourinary system
75350	BPA	Extrophy of urinary bladder	Other anomalies of kidney and collecting system	Genitourinary system
75380	BPA	Absence of bladder or urethra	Other anomalies of kidney and collecting system	Genitourinary system
75385	BPA	Ectopic urethra or urethral orifice	Other anomalies of kidney and collecting system	Genitourinary system
75386	BPA	Fistulae of digestive & urinary tract	Other anomalies of kidney and collecting system	Genitourinary system
75388	BPA	Anomaly of bladder or urethra - other specified NEC	Other anomalies of kidney and collecting system	Genitourinary system
755509	BPA	Anomaly of fingers - major - NOS	Other anomalies of limbs	Musculoskeletal system
75551	BPA	Anomaly of hand	Other anomalies of limbs	Musculoskeletal system
755550	BPA	Cleidocranial dysostosis	Other anomalies of limbs	Musculoskeletal system
755558	BPA	Anomaly of shoulder - other specified NEC	Other anomalies of limbs	Musculoskeletal system
755601	BPA	Hypoplasia of toes or hallux	Other anomalies of limbs	Musculoskeletal system
75581	BPA	Larsen's syndrome	Other anomalies of limbs	Musculoskeletal system
75583	BPA	Pena Shokeir syndrome	Other anomalies of limbs	Musculoskeletal system
7559	BPA	Anomaly of limb - other specified NEC	Other anomalies of limbs	Musculoskeletal system
74850	BPA	Agensis or aplasia of lung	Other anomalies of lung	Respiratory system
74852	BPA	Sequestration of lung	Other anomalies of lung	Respiratory system
74858	BPA	Dysplasia of lung - other specified NEC & NOS	Other anomalies of lung	Respiratory system
74862	BPA	Accessory lobe of lung	Other anomalies of lung	Respiratory system
74868	BPA	Anomaly of lung - other specified NEC	Other anomalies of lung	Respiratory system
75253	BPA	Ectopic testicle (unilateral or bilateral)	Other anomalies of male genitals	Genitourinary system
75261	BPA	Epispadias	Other anomalies of male genitals	Genitourinary system
752822	BPA	Bifid scrotum	Other anomalies of male genitals	Genitourinary system
752828	BPA	Anomaly of testicle & scrotum - other specified NEC	Other anomalies of male genitals	Genitourinary system
752829	BPA	Anomaly of testicle & scrotum - NOS	Other anomalies of male genitals	Genitourinary system
75285	BPA	Absence or aplasia of penis	Other anomalies of male genitals	Genitourinary system
74358	BPA	Anomaly of posterior segment - other specified NEC	Other anomalies of posterior segment	Eye
74738	BPA	Anomaly of pulmonary artery - other specified NEC	Other anomalies of pulmonary artery	Cardiovascular system
74605	BPA	Hypoplasia or dysplasia of pulmonary valve	Other anomalies of pulmonary valve	Cardiovascular system
74608	BPA	Anomaly of pulmonary valve - other specified NEC	Other anomalies of pulmonary valve	Cardiovascular system
74609	BPA	Anomaly of pulmonary valve - NOS	Other anomalies of pulmonary valve	Cardiovascular system
74462	BPA	Facial cleft - NOS	Other anomalies of skull and jaw (includes face,head,neck)	Ear, face and neck
74463	BPA	Hypoplasia - mid-face	Other anomalies of skull and jaw (includes face,head,neck)	Ear, face and neck
74480	BPA	Macrostomia	Other anomalies of skull and jaw (includes face,head,neck)	Ear, face and neck
74488	BPA	Anomaly of face - other specified NEC	Other anomalies of skull and jaw (includes face,head,neck)	Ear, face and neck
756088	BPA	Anomaly of skull & face bones - other specified NEC	Other anomalies of skull and jaw (includes face,head,neck)	Ear, face and neck
74252	BPA	Diastematomyelia	Other anomalies of spinal cord and nervous system	Nervous system
74255	BPA	Syringomyelia	Other anomalies of spinal cord and nervous system	Nervous system
74256	BPA	Tethered Spinal Cord NOS	Other anomalies of spinal cord and nervous system	Nervous system
74258	BPA	Anomaly of spinal cord & meninges - other specified NEC	Other anomalies of spinal cord and nervous system	Nervous system
74288	BPA	Anomalies of nervous system - other specified NEC	Other anomalies of spinal cord and nervous system	Nervous system
74291	BPA	Anomaly of spinal cord - NOS	Other anomalies of spinal cord and nervous system	Nervous system
74299	BPA	Anomaly of nervous system - NOS	Other anomalies of spinal cord and nervous system	Nervous system

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APPENDIX 4: (Continued)

Code	Type	Description	Specific condition group	Major diagnostic category
75901	BPA	Hypoplasia of spleen	Other anomalies of spleen	Other and unspecified anomalies
759041	BPA	Polysplenia syndrome - bilateral left sided sequence	Other anomalies of spleen	Other and unspecified anomalies
75908	BPA	Anomaly of spleen - other specified NEC	Other anomalies of spleen	Other and unspecified anomalies
747610	BPA	Absence of renal artery	Other anomalies of the cardiovascular system	Cardiovascular system
74762	BPA	Arteriovenous malformation - peripheral	Other anomalies of the cardiovascular system	Cardiovascular system
74764	BPA	Anomaly of peripheral arteries - other specified NEC	Other anomalies of the cardiovascular system	Cardiovascular system
74765	BPA	Anomaly of peripheral veins - other specified NEC	Other anomalies of the cardiovascular system	Cardiovascular system
74768	BPA	Anomaly of peripheral vascular system - other specified NEC	Other anomalies of the cardiovascular system	Cardiovascular system
74780	BPA	Arteriovenous aneurysm - brain/cerebral vessels	Other anomalies of the cardiovascular system	Cardiovascular system
75719	BPA	Ichthyosis congenita - other specified NEC & NOS	Other anomalies of the integument	Integumentary system
75730	BPA	Syndromes involving skin anomalies - NEC	Other anomalies of the integument	Integumentary system
75733	BPA	Epidermolysis bullosa	Other anomalies of the integument	Integumentary system
75735	BPA	Incontinentia pigmenti	Other anomalies of the integument	Integumentary system
75737	BPA	Cutis laxa hyperelastica	Other anomalies of the integument	Integumentary system
757384	BPA	Naevus - vascular - NOS	Other anomalies of the integument	Integumentary system
757391	BPA	Cutis aplasia congenita	Other anomalies of the integument	Integumentary system
75740	BPA	Alopecia	Other anomalies of the integument	Integumentary system
75960	BPA	Peutz Jegher syndrome	Other anomalies of the integument	Integumentary system
75961	BPA	Encephalocutaneous angiomatosis	Other anomalies of the integument	Integumentary system
75968	BPA	Hamartosis - other specified NEC	Other anomalies of the integument	Integumentary system
74618	BPA	Anomaly of tricuspid valve - other specified NEC	Other anomalies of tricuspid valve	Cardiovascular system
75854	BPA	Translocations - other specified NEC	Other chromosomal anomalies	Chromosomal anomalies
75855	BPA	Additional marker autosome	Other chromosomal anomalies	Chromosomal anomalies
75858	BPA	Anomaly of autosome - other specified NEC	Other chromosomal anomalies	Chromosomal anomalies
75890	BPA	Mosaicism - NOS	Other chromosomal anomalies	Chromosomal anomalies
75893	BPA	Duplication of chromosome - NOS	Other chromosomal anomalies	Chromosomal anomalies
75898	BPA	Anomaly of chromosome - other specified NEC	Other chromosomal anomalies	Chromosomal anomalies
75899	BPA	Anomaly of chromosome - NOS	Other chromosomal anomalies	Chromosomal anomalies
75948	BPA	Conjoined twins - other specified NEC	Other congenital anomalies	Other and unspecified anomalies
75970	BPA	Acardiac monster	Other congenital anomalies	Other and unspecified anomalies
75991	BPA	Embryopathia - other specified NEC	Other congenital anomalies	Other and unspecified anomalies
75999	BPA	Congenital anomaly - NOS	Other congenital anomalies	Other and unspecified anomalies
75604	BPA	Mandibulofacial dysostosis	Other congenital malformation syndromes	Congenital malformation syndromes
75606	BPA	Goldenhar's syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
759242	BPA	Di George syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
759803	BPA	Waardenburg syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
759804	BPA	Saethre Chotzen syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
759807	BPA	Williams syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
759808	BPA	Syndromes Affecting Facial Appearance - Other	Other congenital malformation syndromes	Congenital malformation syndromes
759811	BPA	Amniotic band association	Other congenital malformation syndromes	Congenital malformation syndromes
759821	BPA	Cornelia de Lange syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
759823	BPA	Laurence Moon Biedel syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
759826	BPA	Smith Lemli Opitz syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
759843	BPA	Klippel Trenaunay Weber syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
759846	BPA	Caudal regression syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
759865	BPA	Other conditions NEC - Pentalogy of Cantrell	Other congenital malformation syndromes	Congenital malformation syndromes
759872	BPA	Beckwith syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
759874	BPA	Prader Willi syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
759894	BPA	Meckel Gruber syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
759898	BPA	Syndrome - other specified NEC	Other congenital malformation syndromes	Congenital malformation syndromes
759899	BPA	Syndrome - unidentified - NOS	Other congenital malformation syndromes	Congenital malformation syndromes
75028	BPA	Anomaly of mouth & pharynx - other specified NEC	Other gastrointestinal anomalies	Gastrointestinal system
75034	BPA	Stenosis or stricture of oesophagus	Other gastrointestinal anomalies	Gastrointestinal system
75070	BPA	Microgastria	Other gastrointestinal anomalies	Gastrointestinal system
75073	BPA	Displacement or transposition of stomach	Other gastrointestinal anomalies	Gastrointestinal system
75078	BPA	Anomaly of stomach - other specified NEC	Other gastrointestinal anomalies	Gastrointestinal system
75141	BPA	Anomaly of mesentery	Other gastrointestinal anomalies	Gastrointestinal system
75154	BPA	Anal fistula	Other gastrointestinal anomalies	Gastrointestinal system
75188	BPA	Anomaly of digestive system - other specified NEC	Other gastrointestinal anomalies	Gastrointestinal system
75242	BPA	Rectovaginal fistula	Other gastrointestinal anomalies	Gastrointestinal system
7459	BPA	Anomaly of cardiac septal closure - NOS	Other malformations cardiac chambers and connections	Cardiovascular system

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APPENDIX 4: (Continued)

Code	Type	Description	Specific condition group	Major diagnostic category
75480	BPA	Pigeon chest	Other musculoskeletal anomalies	Musculoskeletal system
75481	BPA	Funnel chest	Other musculoskeletal anomalies	Musculoskeletal system
755670	BPA	Absence of pelvis	Other musculoskeletal anomalies	Musculoskeletal system
75680	BPA	Poland syndrome	Other musculoskeletal anomalies	Musculoskeletal system
753211	BPA	Stenosis or stricture of ureter	Other obstructive anomalies of renal pelvis and ureter	Genitourinary system
742270	BPA	Absence of septum pellucidum - without mention of septo-optic dysplasia	Other reduction deformities of brain	Nervous system
742271	BPA	Absence of septum pellucidum - with septo-optic dysplasia	Other reduction deformities of brain	Nervous system
74228	BPA	Reduction deformity of brain - other specified NEC	Other reduction deformities of brain	Nervous system
74383	BPA	Septo-optic dysplasia	Other reduction deformities of brain	Nervous system
75323	BPA	Pelvic-ureteric junction obstruction	Pelvic-ureteric junction obstruction	Genitourinary system
2701	BPA	Phenylketonuria	Phenylketonuria	Other and unspecified anomalies
75603	BPA	Pierre Robin syndrome	Pierre Robin syndrome	Congenital malformation syndromes
75500	BPA	Polydactyly - accessory fingers	Polydactyly	Musculoskeletal system
75501	BPA	Polydactyly - accessory thumbs	Polydactyly	Musculoskeletal system
75502	BPA	Polydactyly - accessory toes	Polydactyly	Musculoskeletal system
75503	BPA	Polydactyly - accessory hallux	Polydactyly	Musculoskeletal system
75507	BPA	Polydactyly - accessory digits - feet NOS	Polydactyly	Musculoskeletal system
75508	BPA	Polydactyly - accessory digits - hand NOS	Polydactyly	Musculoskeletal system
75509	BPA	Polydactyly - accessory digits - NOS	Polydactyly	Musculoskeletal system
75360	BPA	Posterior urethral valves	Posterior urethral valves	Genitourinary system
755230	BPA	Reduction of limb - total absence of forearm - hand absent	Reduction deformities of limbs	Musculoskeletal system
755239	BPA	Reduction of limb - absence of forearm NOS - hand absent	Reduction deformities of limbs	Musculoskeletal system
755240	BPA	Reduction of limb - total absence of hand & digits	Reduction deformities of limbs	Musculoskeletal system
755242	BPA	Reduction of limb - total absence of thumb	Reduction deformities of limbs	Musculoskeletal system
755243	BPA	Reduction of limb - total absence of 2nd &/or 3rd fingers	Reduction deformities of limbs	Musculoskeletal system
755244	BPA	Reduction of limb - total absence of 4th &/or 5th fingers	Reduction deformities of limbs	Musculoskeletal system
755245	BPA	Reduction of limb - other total absence of fingers NEC	Reduction deformities of limbs	Musculoskeletal system
755246	BPA	Reduction of limb - partial absence of thumb	Reduction deformities of limbs	Musculoskeletal system
755247	BPA	Reduction of limb - partial absence of fingers	Reduction deformities of limbs	Musculoskeletal system
755248	BPA	Reduction of limb - absence of hand &/or fingers - other specified NEC	Reduction deformities of limbs	Musculoskeletal system
755249	BPA	Reduction of limb - absence of hand &/or fingers - NOS	Reduction deformities of limbs	Musculoskeletal system
755251	BPA	Short or hypoplastic forearm - NOS	Reduction deformities of limbs	Musculoskeletal system
755259	BPA	Short or hypoplastic arm - NOS	Reduction deformities of limbs	Musculoskeletal system
755260	BPA	Reduction of limb - preaxial - arm - absence of radius	Reduction deformities of limbs	Musculoskeletal system
755261	BPA	Reduction of limb - preaxial - arm - absence of radius & thumb	Reduction deformities of limbs	Musculoskeletal system
755262	BPA	Reduction of limb - preaxial - arm - absence of radius - with hypoplastic thumb	Reduction deformities of limbs	Musculoskeletal system
755263	BPA	Reduction of limb - preaxial - arm - short or hypoplastic radius - NOS	Reduction deformities of limbs	Musculoskeletal system
755269	BPA	Reduction of limb - preaxial - arm - NOS	Reduction deformities of limbs	Musculoskeletal system
755273	BPA	Reduction of limb - postaxial - arm - short or hypoplastic ulna - NOS	Reduction deformities of limbs	Musculoskeletal system
755278	BPA	Reduction of limb - postaxial - arm - other specified NEC	Reduction deformities of limbs	Musculoskeletal system
755279	BPA	Reduction of limb - postaxial - arm - NOS	Reduction deformities of limbs	Musculoskeletal system
75528	BPA	Reduction deformity of upper limb - other specified NEC	Reduction deformities of limbs	Musculoskeletal system
75530	BPA	Reduction of limb - total absence of lower limb	Reduction deformities of limbs	Musculoskeletal system
755343	BPA	Reduction of limb - total absence of 2nd &/or 3rd toes	Reduction deformities of limbs	Musculoskeletal system
755344	BPA	Reduction of limb - total absence of 4th &/or 5th toes	Reduction deformities of limbs	Musculoskeletal system
755345	BPA	Reduction of limb - other total absence of toes NEC	Reduction deformities of limbs	Musculoskeletal system
755346	BPA	Reduction of limb - partial absence of hallux	Reduction deformities of limbs	Musculoskeletal system
755347	BPA	Reduction of limb - partial absence of toes	Reduction deformities of limbs	Musculoskeletal system
755348	BPA	Reduction of limb - absence of foot &/or toes - other specified NEC	Reduction deformities of limbs	Musculoskeletal system
755350	BPA	Short or hypoplastic upper leg - NOS	Reduction deformities of limbs	Musculoskeletal system
755351	BPA	Short or hypoplastic lower leg - NOS	Reduction deformities of limbs	Musculoskeletal system
755352	BPA	Hypoplasia of foot NOS	Reduction deformities of limbs	Musculoskeletal system
755359	BPA	Short or hypoplastic leg - NOS	Reduction deformities of limbs	Musculoskeletal system
755360	BPA	Reduction of limb - preaxial - leg - absence of tibia	Reduction deformities of limbs	Musculoskeletal system
755363	BPA	Reduction of limb - preaxial - leg - short or hypoplastic tibia - NOS	Reduction deformities of limbs	Musculoskeletal system
755369	BPA	Reduction of limb - preaxial - leg - NOS	Reduction deformities of limbs	Musculoskeletal system
755370	BPA	Reduction of limb - postaxial - leg - absence of fibula	Reduction deformities of limbs	Musculoskeletal system
755371	BPA	Reduction of limb - postaxial - leg - absence of fibula & toes	Reduction deformities of limbs	Musculoskeletal system

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APPENDIX 4: (Continued)

Code	Type	Description	Specific condition group	Major diagnostic category
755379	BPA	Reduction of limb - postaxial - leg - NOS	Reduction deformities of limbs	Musculoskeletal system
75538	BPA	Reduction deformity of lower limb - other specified NEC	Reduction deformities of limbs	Musculoskeletal system
755501	BPA	Hypoplasia of fingers or thumb	Reduction deformities of limbs	Musculoskeletal system
75571	BPA	Constriction ring - hand or fingers	Reduction deformities of limbs	Musculoskeletal system
75572	BPA	Constriction ring - upper or lower leg	Reduction deformities of limbs	Musculoskeletal system
75573	BPA	Constriction ring - foot or toes	Reduction deformities of limbs	Musculoskeletal system
75578	BPA	Constriction ring - other specified site NEC	Reduction deformities of limbs	Musculoskeletal system
75579	BPA	Constriction ring - site NOS	Reduction deformities of limbs	Musculoskeletal system
742230	BPA	Absence or agenesis of cerebellum	Reduction deformity of cerebellum	Nervous system
742231	BPA	Hypoplasia of cerebellum	Reduction deformity of cerebellum	Nervous system
742232	BPA	Dysplasia of cerebellum	Reduction deformity of cerebellum	Nervous system
742238	BPA	Reduction deformity of cerebellum- other NEC	Reduction deformity of cerebellum	Nervous system
742239	BPA	Reduction deformity of cerebellum - NOS	Reduction deformity of cerebellum	Nervous system
742210	BPA	Absence or agenesis of corpus callosum	Reduction deformity of corpus callosum	Nervous system
742211	BPA	Hypoplasia of corpus callosum	Reduction deformity of corpus callosum	Nervous system
742212	BPA	Dysplasia of corpus callosum	Reduction deformity of corpus callosum	Nervous system
742218	BPA	Reduction deformity of corpus callosum - other NEC	Reduction deformity of corpus callosum	Nervous system
753000	BPA	Absence or agenesis of kidneys - bilateral	Renal agenesis - dysgenesis	Genitourinary system
753001	BPA	Dysplasia of kidneys - bilateral	Renal agenesis - dysgenesis	Genitourinary system
753002	BPA	Hypoplasia of kidneys - bilateral	Renal agenesis - dysgenesis	Genitourinary system
753009	BPA	Potter syndrome - NOS	Renal agenesis - dysgenesis	Genitourinary system
753010	BPA	Absence or agenesis of kidney - unilateral	Renal agenesis - dysgenesis	Genitourinary system
753011	BPA	Dysplasia of kidney - unilateral	Renal agenesis - dysgenesis	Genitourinary system
753012	BPA	Hypoplasia of kidney - unilateral	Renal agenesis - dysgenesis	Genitourinary system
75309	BPA	Renal agenesis NOS	Renal agenesis - dysgenesis	Genitourinary system
75870	BPA	Klinefelter phenotype - 47XXY	Sex chromosome anomalies	Chromosomal anomalies
75871	BPA	Klinefelter phenotype - variant karyotype (additional X chromosomes)	Sex chromosome anomalies	Chromosomal anomalies
75882	BPA	Mosaic karyotype - XY/XXY	Sex chromosome anomalies	Chromosomal anomalies
75884	BPA	XXX male	Sex chromosome anomalies	Chromosomal anomalies
75885	BPA	XXX female	Sex chromosome anomalies	Chromosomal anomalies
75888	BPA	Anomaly of sex chromosomes - other specified NEC	Sex chromosome anomalies	Chromosomal anomalies
75930	BPA	Situs inversus (complete) with dextrocardia	Situs Inversus	Situs inversus
75933	BPA	Situs inversus abdominis	Situs Inversus	Situs inversus
75939	BPA	Situs inversus - NOS	Situs Inversus	Situs inversus
741005	BPA	Spina bifida - aperta - with hydrocephalus - lumbosacral	Spina Bifida	Nervous system
741009	BPA	Spina bifida - aperta - with hydrocephalus - site NOS	Spina Bifida	Nervous system
741010	BPA	Arnold Chiari malformation NOS	Spina Bifida	Nervous system
741013	BPA	Spina bifida - cystica - with hydrocephalus - Arnold Chiari malformation - thoracolumbar	Spina Bifida	Nervous system
741014	BPA	Spina bifida - cystica - with hydrocephalus - Arnold Chiari malformation - lumbar	Spina Bifida	Nervous system
741015	BPA	Spina bifida - cystica - with hydrocephalus - Arnold Chiari malformation - lumbosacral	Spina Bifida	Nervous system
741016	BPA	Spina bifida - cystica - with hydrocephalus - Arnold Chiari malformation - sacral	Spina Bifida	Nervous system
741017	BPA	Spina bifida - cystica - with hydrocephalus - Arnold Chiari malformation - multiple sites	Spina Bifida	Nervous system
741019	BPA	Spina bifida - cystica - with hydrocephalus - Arnold Chiari malformation - site NOS	Spina Bifida	Nervous system
741051	BPA	Spina bifida - cystica - with hydrocephalus NOS - thoracolumbar	Spina Bifida	Nervous system
741052	BPA	Spina bifida - cystica - with hydrocephalus NOS - lumbosacral	Spina Bifida	Nervous system
74109	BPA	Spina bifida - cystica - with hydrocephalus NOS - site NOS	Spina Bifida	Nervous system
741901	BPA	Spina bifida - aperta - cervical	Spina Bifida	Nervous system
741909	BPA	Spina bifida - aperta - site NOS	Spina Bifida	Nervous system
74192	BPA	Spina bifida - cystica - thoracic	Spina Bifida	Nervous system
741930	BPA	Spina bifida - cystica - lumbar NOS	Spina Bifida	Nervous system
741931	BPA	Spina bifida - cystica - thoracolumbar	Spina Bifida	Nervous system
741932	BPA	Spina bifida - cystica - lumbosacral	Spina Bifida	Nervous system
74194	BPA	Spina bifida - cystica - sacral	Spina Bifida	Nervous system
74198	BPA	Spina bifida - other NEC	Spina Bifida	Nervous system
74199	BPA	Spina bifida - NOS	Spina Bifida	Nervous system
74631	BPA	Stenosis of aortic valve	Stenosis of aortic valve	Cardiovascular system
74732	BPA	Stenosis of pulmonary artery	Stenosis of pulmonary artery	Cardiovascular system
74601	BPA	Stenosis of pulmonary valve	Stenosis of pulmonary valve	Cardiovascular system
75510	BPA	Syndactyly - fingers - fused	Syndactyly	Musculoskeletal system
75512	BPA	Syndactyly - toes - fused	Syndactyly	Musculoskeletal system
755140	BPA	Syndactyly NOS - hands - 3rd & 4th fingers	Syndactyly	Musculoskeletal system

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APPENDIX 4: (Continued)

Code	Type	Description	Specific condition group	Major diagnostic category
755141	BPA	Syndactyly NOS - hands - other specified fingers	Syndactyly	Musculoskeletal system
755150	BPA	Syndactyly NOS - toes - 2nd & 3rd toes	Syndactyly	Musculoskeletal system
755151	BPA	Syndactyly NOS - toes - other specified toes	Syndactyly	Musculoskeletal system
75519	BPA	Syndactyly - NOS	Syndactyly	Musculoskeletal system
75450	BPA	Talipes equinovarus	Talipes	Musculoskeletal system
75460	BPA	Talipes calcaneovalgus	Talipes	Musculoskeletal system
75470	BPA	Pes cavus	Talipes	Musculoskeletal system
75473	BPA	Talipes - NOS	Talipes	Musculoskeletal system
74520	BPA	Tetralogy of Fallot	Tetralogy of Fallot	Cardiovascular system
74521	BPA	Pentalogy of Fallot	Tetralogy of Fallot	Cardiovascular system
28240	BPA	Thalassaemia - Alpha	Thalassaemia Alpha	Other and unspecified anomalies
28244	BPA	Thalassaemia - Beta major	Thalassaemia Beta Major	Other and unspecified anomalies
74742	BPA	Total anomalous pulmonary venous return	Total or partial anomalous pulmonary venous return	Cardiovascular system
747430	BPA	Partial anomalous pulmonary venous return - NOS	Total or partial anomalous pulmonary venous return	Cardiovascular system
75032	BPA	Tracheo-oesophageal fistula - no mention of atresia	Tracheo-oesophageal fistula (TOF) only	Gastrointestinal system
74510	BPA	Transposition of great vessels - complete	Transposition of great vessels	Cardiovascular system
74511	BPA	Transposition of great vessels - incomplete	Transposition of great vessels	Cardiovascular system
74512	BPA	Transposition of great vessels - corrected	Transposition of great vessels	Cardiovascular system
74513	BPA	Double outlet right ventricle	Transposition of great vessels	Cardiovascular system
74515	BPA	D-type transposition of great vessels	Transposition of great vessels	Cardiovascular system
74516	BPA	L-type transposition of great vessels	Transposition of great vessels	Cardiovascular system
74519	BPA	Transposition of great vessels - NOS	Transposition of great vessels	Cardiovascular system
75851	BPA	Trisomy C syndromes - other specified NEC	Trisomies and partial trisomies - remainder	Chromosomal anomalies
75852	BPA	Trisomy syndromes - total - other specified NEC	Trisomies and partial trisomies - remainder	Chromosomal anomalies
75853	BPA	Trisomy syndromes - partial	Trisomies and partial trisomies - remainder	Chromosomal anomalies
75810	BPA	Patau syndrome - trisomy 13	Trisomy 13	Chromosomal anomalies
75812	BPA	Patau syndrome - translocation trisomy 13	Trisomy 13	Chromosomal anomalies
75820	BPA	Edward syndrome - trisomy 18	Trisomy 18	Chromosomal anomalies
75824	BPA	Edward syndrome - mosaic - trisomy 18	Trisomy 18	Chromosomal anomalies
75829	BPA	Edward syndrome - NOS	Trisomy 18	Chromosomal anomalies
75800	BPA	Down syndrome - trisomy 21	Trisomy 21	Chromosomal anomalies
75804	BPA	Down syndrome - mosaic - trisomy 21	Trisomy 21	Chromosomal anomalies
75809	BPA	Down syndrome NOS	Trisomy 21	Chromosomal anomalies
7595	BPA	Tuberous sclerosis	Tuberous sclerosis	Integumentary system
75860	BPA	Turner phenotype - 45X	Turner syndrome	Chromosomal anomalies
758615	BPA	Turner phenotype - other mosaic karyotype - 45X/46XY	Turner syndrome	Chromosomal anomalies
75250	BPA	Undescended testicle - unilateral	Undescended testis	Genitourinary system
75251	BPA	Undescended testicle - bilateral	Undescended testis	Genitourinary system
75252	BPA	Undescended testicle - NOS	Undescended testis	Genitourinary system
75431	BPA	Unstable hip	Unstable hip	Musculoskeletal system
75324	BPA	Ureterocoloe	Ureterocoloe	Genitourinary system
759897	BPA	VATER or VACTERL association	VATER association	Congenital malformation syndromes
74543	BPA	Ventricular septal defect - perimembranous	Ventricular septal defect	Cardiovascular system
74544	BPA	Ventricular septal defect - muscular	Ventricular septal defect	Cardiovascular system
74545	BPA	Ventricular septal defect - subaortic	Ventricular septal defect	Cardiovascular system
74548	BPA	Ventricular septal defect - other specified NEC	Ventricular septal defect	Cardiovascular system
74549	BPA	Ventricular septal defect - NOS	Ventricular septal defect	Cardiovascular system
Q16.1	ICD10AM	Congenital absence, atresia and stricture of auditory canal (external)	Absence-stricture auditory canal	Ear, face and neck
Q33.01	ICD10AM	Congenital cystic adenomatoid lung	Adenomatoid malformation of lung	Respiratory system
Q43.32	ICD10AM	Congenital intra-abdominal adhesions (bands)	Adhesions or bands of omentum & peritoneum	Gastrointestinal system
Q04.34	ICD10AM	Agyria and lissencephaly	Agyria and microgyria	Nervous system
Q04.35	ICD10AM	Microgyria and pachygyria	Agyria and microgyria	Nervous system
Q00.09	ICD10AM	Other anencephaly	Anencephaly	Nervous system
Q14.3	ICD10AM	Congenital malformation of choroid	Anomalies of choroid	Eye
Q14.2	ICD10AM	Congenital malformation of optic disc	Anomalies of optic disc	Eye
Q14.1	ICD10AM	Congenital malformation of retina	Anomalies of retina	Eye
Q76.61	ICD10AM	Congenital absence of rib	Anomalies of ribs and sternum	Musculoskeletal system
Q76.62	ICD10AM	Congenital fusion of ribs	Anomalies of ribs and sternum	Musculoskeletal system
Q76.69	ICD10AM	Other congenital malformation of ribs	Anomalies of ribs and sternum	Musculoskeletal system
Q76.71	ICD10AM	Congenital absence of sternum	Anomalies of ribs and sternum	Musculoskeletal system
Q76.79	ICD10AM	Other specified congenital malformation of sternum	Anomalies of ribs and sternum	Musculoskeletal system

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APPENDIX 4: (Continued)

Code	Type	Description	Specific condition group	Major diagnostic category
Q76.39	ICD10AM	Congenital scoliosis due to other specified congenital bony malformation	Anomalies of spinal vertebrae	Musculoskeletal system
Q76.42	ICD10AM	Congenital anomalies of sacral vertebra(e)	Anomalies of spinal vertebrae	Musculoskeletal system
Q76.43	ICD10AM	Other congenital anomalies of vertebra(e)	Anomalies of spinal vertebrae	Musculoskeletal system
Q76.45	ICD10AM	Hemivertebra	Anomalies of spinal vertebrae	Musculoskeletal system
Q14.0	ICD10AM	Congenital malformation of vitreous humour	Anomalies of vitreous humour	Eye
Q11.1	ICD10AM	Other anophthalmos	Anophthalmos-microphthalmos	Eye
Q11.2	ICD10AM	Microphthalmos	Anophthalmos-microphthalmos	Eye
Q55.00	ICD10AM	Absence and aplasia of testis, unspecified	Aplasia or hypoplasia of testes or scrotum	Genitourinary system
Q55.01	ICD10AM	Absence and aplasia of testis, unilateral	Aplasia or hypoplasia of testes or scrotum	Genitourinary system
Q55.02	ICD10AM	Absence and aplasia of testis, bilateral	Aplasia or hypoplasia of testes or scrotum	Genitourinary system
Q55.1	ICD10AM	Hypoplasia of testis and scrotum	Aplasia or hypoplasia of testes or scrotum	Genitourinary system
Q74.3	ICD10AM	Arthrogryposis multiplex congenita	Arthrogryposis	Musculoskeletal system
Q89.01	ICD10AM	Congenital asplenia	Asplenia	Other and unspecified anomalies
Q23.02	ICD10AM	Congenital atresia of aortic valve	Atresia of aortic valve	Cardiovascular system
Q44.2	ICD10AM	Atresia of bile ducts	Atresia of hepatic or bile ducts	Gastrointestinal system
Q44.71	ICD10AM	Alagille syndrome	Atresia of hepatic or bile ducts	Gastrointestinal system
Q25.5	ICD10AM	Atresia of pulmonary artery	Atresia of pulmonary artery	Cardiovascular system
Q22.0	ICD10AM	Pulmonary valve atresia	Atresia of pulmonary valve	Cardiovascular system
Q22.42	ICD10AM	Congenital tricuspid atresia	Atresia of tricuspid valve	Cardiovascular system
Q42.20	ICD10AM	Congenital absence, atresia and stenosis of anus with unspecified fistula	Atresia-stenosis of anus (includes with fistula)	Gastrointestinal system
Q42.21	ICD10AM	Congenital absence, atresia and stenosis of anus with anocutaneous fistula	Atresia-stenosis of anus (includes with fistula)	Gastrointestinal system
Q42.22	ICD10AM	Congenital absence, atresia and stenosis of anus with anovestibular fistula	Atresia-stenosis of anus (includes with fistula)	Gastrointestinal system
Q42.29	ICD10AM	Congenital absence, atresia and stenosis of anus with other fistula	Atresia-stenosis of anus (includes with fistula)	Gastrointestinal system
Q42.3	ICD10AM	Congenital absence, atresia and stenosis of anus without fistula	Atresia-stenosis of anus (includes with fistula)	Gastrointestinal system
Q42.1	ICD10AM	Congenital absence, atresia and stenosis of rectum without fistula	Atresia-stenosis of large intestine (includes with fistula)	Gastrointestinal system
Q42.8	ICD10AM	Congenital absence, atresia and stenosis of other parts of large intestine	Atresia-stenosis of large intestine (includes with fistula)	Gastrointestinal system
Q42.9	ICD10AM	Congenital absence, atresia and stenosis of large intestine, part unspecified	Atresia-stenosis of large intestine (includes with fistula)	Gastrointestinal system
Q41.0	ICD10AM	Congenital absence, atresia and stenosis of duodenum	Atresia-stenosis of small intestine (includes with fistula)	Gastrointestinal system
Q41.01	ICD10AM	Congenital absence and atresia of duodenum	Atresia-stenosis of small intestine (includes with fistula)	Gastrointestinal system
Q41.1	ICD10AM	Congenital absence, atresia and stenosis of jejunum	Atresia-stenosis of small intestine (includes with fistula)	Gastrointestinal system
Q41.2	ICD10AM	Congenital absence, atresia and stenosis of ileum	Atresia-stenosis of small intestine (includes with fistula)	Gastrointestinal system
Q41.9	ICD10AM	Congenital absence, atresia and stenosis of small intestine, part unspecified	Atresia-stenosis of small intestine (includes with fistula)	Gastrointestinal system
Q21.10	ICD10AM	Atrial septal defect, unspecified	Atrial septal defect	Cardiovascular system
Q21.11	ICD10AM	Patent or persistent foramen ovale	Atrial septal defect	Cardiovascular system
Q21.12	ICD10AM	Sinus venosus defect	Atrial septal defect	Cardiovascular system
Q21.19	ICD10AM	Other specified atrial septal defect	Atrial septal defect	Cardiovascular system
Q21.8	ICD10AM	Other congenital malformations of cardiac septa	Atrial septal defect	Cardiovascular system
Q21.9	ICD10AM	Congenital malformation of cardiac septum, unspecified	Atrial septal defect	Cardiovascular system
Q21.2	ICD10AM	Atrioventricular septal defect	Atrioventricular septal defect	Cardiovascular system
Q21.20	ICD10AM	Atrioventricular septal defect, unspecified	Atrioventricular septal defect	Cardiovascular system
Q93.3	ICD10AM	Deletion of short arm of chromosome 4	Autosomal chromosome deletions	Chromosomal anomalies
Q93.4	ICD10AM	Deletion of short arm of chromosome 5	Autosomal chromosome deletions	Chromosomal anomalies
Q93.5	ICD10AM	Other deletions of part of a chromosome	Autosomal chromosome deletions	Chromosomal anomalies
Q93.8	ICD10AM	Other deletions from the autosomes	Autosomal chromosome deletions	Chromosomal anomalies
Q15.0	ICD10AM	Congenital glaucoma	Buphthalmos-congenital glaucoma	Eye
Q87.86	ICD10AM	CHARGE syndrome	CHARGE syndrome	Congenital malformation syndromes
Q30.0	ICD10AM	Choanal atresia and stenosis	Choanal atresia and stenosis	Respiratory system
Q30.02	ICD10AM	Choanal stenosis	Choanal atresia and stenosis	Respiratory system
Q77.1	ICD10AM	Thanatophoric short stature	Chondrodystrophy	Musculoskeletal system
Q77.2	ICD10AM	Short rib syndrome	Chondrodystrophy	Musculoskeletal system
Q77.4	ICD10AM	Achondroplasia	Chondrodystrophy	Musculoskeletal system
Q77.7	ICD10AM	Spondyloepiphyseal dysplasia	Chondrodystrophy	Musculoskeletal system
Q37.0	ICD10AM	Cleft hard palate with bilateral cleft lip	Cleft lip and palate	Gastrointestinal system
Q37.1	ICD10AM	Cleft hard palate with unilateral cleft lip	Cleft lip and palate	Gastrointestinal system
Q37.2	ICD10AM	Cleft soft palate with bilateral cleft lip	Cleft lip and palate	Gastrointestinal system
Q37.3	ICD10AM	Cleft soft palate with unilateral cleft lip	Cleft lip and palate	Gastrointestinal system
Q37.4	ICD10AM	Cleft hard and soft palate with bilateral cleft lip	Cleft lip and palate	Gastrointestinal system
Q37.5	ICD10AM	Cleft hard and soft palate with unilateral cleft lip	Cleft lip and palate	Gastrointestinal system
Q37.8	ICD10AM	Unspecified cleft palate with bilateral cleft lip	Cleft lip and palate	Gastrointestinal system
Q37.9	ICD10AM	Unspecified cleft palate with unilateral cleft lip	Cleft lip and palate	Gastrointestinal system
Q36.0	ICD10AM	Cleft lip, bilateral	Cleft lip only	Gastrointestinal system

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APPENDIX 4: (Continued)

Code	Type	Description	Specific condition group	Major diagnostic category
Q36.1	ICD10AM	Cleft lip, median	Cleft lip only	Gastrointestinal system
Q36.9	ICD10AM	Cleft lip, unilateral	Cleft lip only	Gastrointestinal system
Q35.1	ICD10AM	Cleft hard palate	Cleft palate only	Gastrointestinal system
Q35.11	ICD10AM	Cleft hard palate, bilateral	Cleft palate only	Gastrointestinal system
Q35.12	ICD10AM	Cleft hard palate, median	Cleft palate only	Gastrointestinal system
Q35.3	ICD10AM	Cleft soft palate	Cleft palate only	Gastrointestinal system
Q35.30	ICD10AM	Cleft soft palate, unspecified	Cleft palate only	Gastrointestinal system
Q35.31	ICD10AM	Cleft soft palate, bilateral	Cleft palate only	Gastrointestinal system
Q35.32	ICD10AM	Cleft soft palate, median	Cleft palate only	Gastrointestinal system
Q35.5	ICD10AM	Cleft hard palate with cleft soft palate	Cleft palate only	Gastrointestinal system
Q35.9	ICD10AM	Cleft palate, unspecified	Cleft palate only	Gastrointestinal system
Q38.5	ICD10AM	Congenital malformations of palate, not elsewhere classified	Cleft palate only	Gastrointestinal system
Q25.1	ICD10AM	Coarctation of aorta	Coarctation of aorta	Cardiovascular system
Q25.10	ICD10AM	Coarctation of aorta, unspecified	Coarctation of aorta	Cardiovascular system
Q13.0	ICD10AM	Coloboma of iris	Coloboma of iris	Eye
Q20.0	ICD10AM	Common arterial trunk	Common arterial trunk	Cardiovascular system
Q21.4	ICD10AM	Aortopulmonary septal defect	Common arterial trunk	Cardiovascular system
Q20.4	ICD10AM	Double inlet ventricle	Common ventricle	Cardiovascular system
Q12.0	ICD10AM	Congenital cataract	Congenital cataract	Eye
Q65.0	ICD10AM	Congenital dislocation of hip, unilateral	Congenital dislocation of hips	Musculoskeletal system
Q65.1	ICD10AM	Congenital dislocation of hip, bilateral	Congenital dislocation of hips	Musculoskeletal system
Q65.2	ICD10AM	Congenital dislocation of hip, unspecified	Congenital dislocation of hips	Musculoskeletal system
Q03.0	ICD10AM	Malformations of aqueduct of Sylvius	Congenital hydrocephalus	Nervous system
Q03.1	ICD10AM	Atresia of foramina of Magendie and Luschka	Congenital hydrocephalus	Nervous system
Q03.8	ICD10AM	Other congenital hydrocephalus	Congenital hydrocephalus	Nervous system
Q03.9	ICD10AM	Congenital hydrocephalus, unspecified	Congenital hydrocephalus	Nervous system
E03.0	ICD10AM	Congenital hypothyroidism with diffuse goitre	Congenital hypothyroidism	Other and unspecified anomalies
E03.1	ICD10AM	Congenital hypothyroidism without goitre	Congenital hypothyroidism	Other and unspecified anomalies
Q75.01	ICD10AM	Coronal craniosynostosis	Craniosynostosis	Musculoskeletal system
Q75.02	ICD10AM	Sagittal craniosynostosis	Craniosynostosis	Musculoskeletal system
Q75.03	ICD10AM	Trigonocephaly	Craniosynostosis	Musculoskeletal system
Q75.04	ICD10AM	Craniosynostosis of other multiple sutures	Craniosynostosis	Musculoskeletal system
Q75.09	ICD10AM	Other and unspecified craniosynostosis	Craniosynostosis	Musculoskeletal system
Q75.1	ICD10AM	Craniofacial dysostosis	Craniosynostosis	Musculoskeletal system
E84.0	ICD10AM	Cystic fibrosis with pulmonary manifestations	Cystic fibrosis	Other and unspecified anomalies
E84.1	ICD10AM	Cystic fibrosis with intestinal manifestations	Cystic fibrosis	Other and unspecified anomalies
E84.8	ICD10AM	Cystic fibrosis with other manifestations	Cystic fibrosis	Other and unspecified anomalies
E84.9	ICD10AM	Cystic fibrosis, unspecified	Cystic fibrosis	Other and unspecified anomalies
Q61.1	ICD10AM	Polycystic kidney, autosomal recessive	Cystic kidney disease	Genitourinary system
Q61.3	ICD10AM	Polycystic kidney, unspecified	Cystic kidney disease	Genitourinary system
Q61.40	ICD10AM	Renal dysplasia, unspecified	Cystic kidney disease	Genitourinary system
Q61.41	ICD10AM	Cystic renal dysplasia, unilateral	Cystic kidney disease	Genitourinary system
Q61.42	ICD10AM	Cystic renal dysplasia, bilateral	Cystic kidney disease	Genitourinary system
Q61.8	ICD10AM	Other cystic kidney diseases	Cystic kidney disease	Genitourinary system
Q24.0	ICD10AM	Dextrocardia	Dextrocardia	Cardiovascular system
Q79.0	ICD10AM	Congenital diaphragmatic hernia	Diaphragmatic hernia	Musculoskeletal system
Q62.51	ICD10AM	Double ureter	Duplex kidney and/or collecting system	Genitourinary system
Q62.59	ICD10AM	Other duplication of ureter	Duplex kidney and/or collecting system	Genitourinary system
Q63.01	ICD10AM	Double kidney	Duplex kidney and/or collecting system	Genitourinary system
Q22.5	ICD10AM	Ebstein's anomaly	Ebstein's anomaly	Cardiovascular system
Q43.5	ICD10AM	Ectopic anus	Ectopic anus	Gastrointestinal system
Q01.0	ICD10AM	Frontal encephalocele	Encephalocele	Nervous system
Q01.2	ICD10AM	Occipital encephalocele	Encephalocele	Nervous system
Q01.81	ICD10AM	Parietal encephalocele	Encephalocele	Nervous system
Q01.89	ICD10AM	Encephalocele of other specified sites	Encephalocele	Nervous system
Q01.9	ICD10AM	Encephalocele, unspecified	Encephalocele	Nervous system
Q79.2	ICD10AM	Exomphalos	Exomphalos	Musculoskeletal system
Q79.3	ICD10AM	Gastroschisis	Gastroschisis	Musculoskeletal system
Q43.10	ICD10AM	Hirschsprung's disease, unspecified	Hirschsprungs Disease	Gastrointestinal system
Q43.11	ICD10AM	Short segment Hirschsprung's disease	Hirschsprungs Disease	Gastrointestinal system
Q43.12	ICD10AM	Long segment Hirschsprung's disease	Hirschsprungs Disease	Gastrointestinal system

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APPENDIX 4: (Continued)

Code	Type	Description	Specific condition group	Major diagnostic category
Q43.19	ICD10AM	Other Hirschsprung's disease	Hirschsprungs Disease	Gastrointestinal system
Q04.2	ICD10AM	Holoprosencephaly	Holoprosencephaly	Nervous system
Q63.11	ICD10AM	Horseshoe kidney	Horseshoe kidney	Genitourinary system
Q62.0	ICD10AM	Congenital hydronephrosis	Hydronephrosis	Genitourinary system
P83.2	ICD10AM	Hydrops fetalis not due to haemolytic disease	Hydrops not due to isoimmunization	Non-immune hydrops foetalis
Q62.2	ICD10AM	Congenital megaloureter	Hydroureter and megaloureter	Genitourinary system
Q62.34	ICD10AM	Congenital hydroureter	Hydroureter and megaloureter	Genitourinary system
Q07.82	ICD10AM	Optic nerve hypoplasia	Hypoplasia of optic nerve	Eye
Q20.81	ICD10AM	Hypoplastic right ventricle	Hypoplastic heart	Cardiovascular system
Q22.6	ICD10AM	Hypoplastic right heart syndrome	Hypoplastic heart	Cardiovascular system
Q23.4	ICD10AM	Hypoplastic left heart syndrome	Hypoplastic heart	Cardiovascular system
Q54.0	ICD10AM	Hypospadias, balanic	Hypospadias	Genitourinary system
Q54.1	ICD10AM	Hypospadias, penile	Hypospadias	Genitourinary system
Q54.2	ICD10AM	Hypospadias, penoscrotal	Hypospadias	Genitourinary system
Q54.3	ICD10AM	Hypospadias, perineal	Hypospadias	Genitourinary system
Q54.8	ICD10AM	Other hypospadias	Hypospadias	Genitourinary system
Q54.9	ICD10AM	Hypospadias, unspecified	Hypospadias	Genitourinary system
Q56.4	ICD10AM	Indeterminate sex and ambiguous genitalia	Indeterminate sex - ambiguous genitalia	Genitourinary system
Q43.31	ICD10AM	Malrotation of colon	Intestinal malrotation	Gastrointestinal system
D18.1	ICD10AM	Lymphangioma, any site	Lymphangioma	Integumentary system
Q02	ICD10AM	Microcephaly	Microcephaly	Nervous system
Q87.13	ICD10AM	Noonan syndrome	Noonan syndrome	Congenital malformation syndromes
Q30.2	ICD10AM	Fissured, notched and cleft nose	Nose, larynx, trachea, bronchus	Respiratory system
Q30.8	ICD10AM	Other congenital malformations of nose	Nose, larynx, trachea, bronchus	Respiratory system
Q30.9	ICD10AM	Congenital malformation of nose, unspecified	Nose, larynx, trachea, bronchus	Respiratory system
Q31.0	ICD10AM	Web of larynx	Nose, larynx, trachea, bronchus	Respiratory system
Q31.1	ICD10AM	Congenital subglottic stenosis	Nose, larynx, trachea, bronchus	Respiratory system
Q31.8	ICD10AM	Other congenital malformations of larynx	Nose, larynx, trachea, bronchus	Respiratory system
Q32.1	ICD10AM	Other congenital malformations of trachea	Nose, larynx, trachea, bronchus	Respiratory system
Q32.4	ICD10AM	Other congenital malformations of bronchus	Nose, larynx, trachea, bronchus	Respiratory system
Q62.8	ICD10AM	Other congenital malformations of ureter	Obstructive anomalies of renal pelvis and ureter	Genitourinary system
Q39.0	ICD10AM	Atresia of oesophagus without fistula	Oesophageal atresia only	Gastrointestinal system
Q39.11	ICD10AM	Atresia of oesophagus with fistula between trachea and upper oesophageal pouch	Oesophageal atresia with TOF	Gastrointestinal system
Q39.12	ICD10AM	Atresia of oesophagus with fistula between trachea and lower oesophageal pouch	Oesophageal atresia with TOF	Gastrointestinal system
Q39.19	ICD10AM	Atresia of oesophagus with tracheo-oesophageal fistula	Oesophageal atresia with TOF	Gastrointestinal system
Q78.0	ICD10AM	Osteogenesis imperfecta	Osteodystrophy	Musculoskeletal system
Q78.8	ICD10AM	Other specified osteochondrodysplasias	Osteodystrophy	Musculoskeletal system
Q78.89	ICD10AM	Other specified osteochondrodysplasias	Osteodystrophy	Musculoskeletal system
Q79.4	ICD10AM	Prune belly syndrome	Other anomalies of abdominal wall	Musculoskeletal system
Q79.5	ICD10AM	Other congenital malformations of abdominal wall	Other anomalies of abdominal wall	Musculoskeletal system
Q89.12	ICD10AM	Congenital adrenal hypoplasia	Other anomalies of adrenal gland	Other and unspecified anomalies
Q89.19	ICD10AM	Other specified congenital malformations of adrenal gland	Other anomalies of adrenal gland	Other and unspecified anomalies
Q13.2	ICD10AM	Other congenital malformations of iris	Other anomalies of anterior segment	Eye
Q13.4	ICD10AM	Other and unspecified congenital corneal malformations	Other anomalies of anterior segment	Eye
Q13.5	ICD10AM	Blue sclera	Other anomalies of anterior segment	Eye
Q13.8	ICD10AM	Other congenital malformations of anterior segment of eye	Other anomalies of anterior segment	Eye
Q13.9	ICD10AM	Congenital malformation of anterior segment of eye, unspecified	Other anomalies of anterior segment	Eye
Q25.2	ICD10AM	Atresia of aorta	Other anomalies of aorta	Cardiovascular system
Q25.3	ICD10AM	Stenosis of aorta	Other anomalies of aorta	Cardiovascular system
Q25.4	ICD10AM	Other and unspecified congenital malformations of aorta	Other anomalies of aorta	Cardiovascular system
Q25.43	ICD10AM	Double aortic arch [vascular ring of aorta]	Other anomalies of aorta	Cardiovascular system
Q23.21	ICD10AM	Congenital mitral stenosis	Other anomalies of aortic and mitral valve	Cardiovascular system
Q23.22	ICD10AM	Congenital mitral atresia	Other anomalies of aortic and mitral valve	Cardiovascular system
Q23.8	ICD10AM	Other congenital malformations of aortic and mitral valves	Other anomalies of aortic and mitral valve	Cardiovascular system
Q23.9	ICD10AM	Congenital malformation of aortic and mitral valves, unspecified	Other anomalies of aortic and mitral valve	Cardiovascular system
Q04.5	ICD10AM	Megalencephaly	Other anomalies of brain	Nervous system
Q04.60	ICD10AM	Congenital cerebral cysts, unspecified	Other anomalies of brain	Nervous system
Q04.8	ICD10AM	Other specified congenital malformations of brain	Other anomalies of brain	Nervous system
Q04.9	ICD10AM	Congenital malformation of brain, unspecified	Other anomalies of brain	Nervous system
Q79.1	ICD10AM	Other and unspecified congenital malformations of diaphragm	Other anomalies of diaphragm	Musculoskeletal system

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APPENDIX 4: (Continued)

Code	Type	Description	Specific condition group	Major diagnostic category
Q16.4	ICD10AM	Other congenital malformations of middle ear	Other anomalies of ear	Ear, face and neck
Q16.5	ICD10AM	Congenital malformation of inner ear	Other anomalies of ear	Ear, face and neck
Q16.9	ICD10AM	Congenital malformation of ear causing impairment of hearing, unspecified	Other anomalies of ear	Ear, face and neck
Q17.8	ICD10AM	Other specified congenital malformations of ear	Other anomalies of ear	Ear, face and neck
Q17.9	ICD10AM	Congenital malformation of ear, unspecified	Other anomalies of ear	Ear, face and neck
Q89.21	ICD10AM	Congenital malformations of pituitary gland	Other anomalies of endocrine glands	Other and unspecified anomalies
Q89.26	ICD10AM	Congenital malformations of thymus	Other anomalies of endocrine glands	Other and unspecified anomalies
Q89.29	ICD10AM	Congenital malformations of other specified endocrine glands	Other anomalies of endocrine glands	Other and unspecified anomalies
Q10.3	ICD10AM	Other congenital malformations of eyelid	Other anomalies of eye	Eye
Q10.4	ICD10AM	Absence and agenesis of lacrimal apparatus	Other anomalies of eye	Eye
Q11.3	ICD10AM	Macrophthalmos	Other anomalies of eye	Eye
Q15.8	ICD10AM	Other specified congenital malformations of eye	Other anomalies of eye	Eye
Q15.9	ICD10AM	Congenital malformation of eye, unspecified	Other anomalies of eye	Eye
Q50.00	ICD10AM	Congenital absence of ovary, unspecified	Other anomalies of female genitals	Genitourinary system
Q51.3	ICD10AM	Bicornuate uterus	Other anomalies of female genitals	Genitourinary system
Q52.2	ICD10AM	Congenital rectovaginal fistula	Other anomalies of female genitals	Genitourinary system
Q52.6	ICD10AM	Other and unspecified congenital malformations of clitoris	Other anomalies of female genitals	Genitourinary system
Q52.7	ICD10AM	Other and unspecified congenital malformations of vulva	Other anomalies of female genitals	Genitourinary system
Q52.8	ICD10AM	Other specified congenital malformations of female genitalia	Other anomalies of female genitals	Genitourinary system
Q52.9	ICD10AM	Congenital malformation of female genitalia, unspecified	Other anomalies of female genitals	Genitourinary system
Q44.1	ICD10AM	Other congenital malformations of gallbladder	Other anomalies of gallbladder, bile duct, pancreas and liver	Gastrointestinal system
Q44.3	ICD10AM	Congenital stenosis and stricture of bile ducts	Other anomalies of gallbladder, bile duct, pancreas and liver	Gastrointestinal system
Q44.5	ICD10AM	Other congenital malformations of bile ducts	Other anomalies of gallbladder, bile duct, pancreas and liver	Gastrointestinal system
Q44.79	ICD10AM	Other congenital malformations of liver	Other anomalies of gallbladder, bile duct, pancreas and liver	Gastrointestinal system
Q45.1	ICD10AM	Annular pancreas	Other anomalies of gallbladder, bile duct, pancreas and liver	Gastrointestinal system
Q45.39	ICD10AM	Other congenital malformations of pancreas and pancreatic duct	Other anomalies of gallbladder, bile duct, pancreas and liver	Gastrointestinal system
Q26.9	ICD10AM	Congenital malformation of great vein, unspecified	Other anomalies of great veins	Cardiovascular system
Q20.89	ICD10AM	Other specified congenital malformations of cardiac chambers and connections	Other anomalies of heart	Cardiovascular system
Q24.1	ICD10AM	Laevocardia	Other anomalies of heart	Cardiovascular system
Q24.2	ICD10AM	Cor triatriatum	Other anomalies of heart	Cardiovascular system
Q24.3	ICD10AM	Pulmonary infundibular stenosis	Other anomalies of heart	Cardiovascular system
Q24.4	ICD10AM	Congenital subaortic stenosis	Other anomalies of heart	Cardiovascular system
Q24.9	ICD10AM	Congenital malformation of heart, unspecified	Other anomalies of heart	Cardiovascular system
Q43.39	ICD10AM	Other congenital malformations of intestinal fixation	Other anomalies of intestinal fixation	Gastrointestinal system
Q62.62	ICD10AM	Malposition of ureter, ureteric drainage via urethra	Other anomalies of kidney and collecting system	Genitourinary system
Q63.9	ICD10AM	Congenital malformation of kidney, unspecified	Other anomalies of kidney and collecting system	Genitourinary system
Q64.19	ICD10AM	Exstrophy of urinary bladder	Other anomalies of kidney and collecting system	Genitourinary system
Q64.31	ICD10AM	Congenital bladder neck obstruction	Other anomalies of kidney and collecting system	Genitourinary system
Q64.32	ICD10AM	Congenital stricture of urethra	Other anomalies of kidney and collecting system	Genitourinary system
Q64.75	ICD10AM	Congenital gastrointestinal-urinary tract fistula	Other anomalies of kidney and collecting system	Genitourinary system
Q64.79	ICD10AM	Other congenital malformations of bladder and urethra	Other anomalies of kidney and collecting system	Genitourinary system
Q74.09	ICD10AM	Other congenital malformations of upper limb(s), including shoulder girdle	Other anomalies of limbs	Musculoskeletal system
Q33.2	ICD10AM	Sequestration of lung	Other anomalies of lung	Respiratory system
Q33.8	ICD10AM	Other congenital malformations of lung	Other anomalies of lung	Respiratory system
Q33.9	ICD10AM	Congenital malformation of lung, unspecified	Other anomalies of lung	Respiratory system
Q34.8	ICD10AM	Other specified congenital malformations of respiratory system	Other anomalies of lung	Respiratory system
Q34.9	ICD10AM	Congenital malformation of respiratory system, unspecified	Other anomalies of lung	Respiratory system
Q53.0	ICD10AM	Ectopic testis	Other anomalies of male genitals	Genitourinary system
Q55.22	ICD10AM	Bifid scrotum	Other anomalies of male genitals	Genitourinary system
Q55.29	ICD10AM	Other congenital malformations of testis or scrotum	Other anomalies of male genitals	Genitourinary system
Q55.4	ICD10AM	Other and unspecified congenital malformations of vas deferens, epididymis, seminal vesicles and prostate	Other anomalies of male genitals	Genitourinary system
Q55.5	ICD10AM	Congenital absence and aplasia of penis	Other anomalies of male genitals	Genitourinary system
Q64.0	ICD10AM	Epispadias	Other anomalies of male genitals	Genitourinary system
Q14.8	ICD10AM	Other congenital malformations of posterior segment of eye	Other anomalies of posterior segment	Eye
Q25.7	ICD10AM	Other and unspecified congenital malformations of pulmonary artery	Other anomalies of pulmonary artery	Cardiovascular system
Q22.3	ICD10AM	Other and unspecified congenital malformations of pulmonary valve	Other anomalies of pulmonary valve	Cardiovascular system
Q67.49	ICD10AM	Other congenital deformities of skull, face and jaw	Other anomalies of skull and jaw (includes face,head,neck)	Musculoskeletal system
Q75.81	ICD10AM	Frontonasal dysplasia	Other anomalies of skull and jaw (includes face,head,neck)	Musculoskeletal system
Q75.89	ICD10AM	Other specified congenital malformations of skull and face bones	Other anomalies of skull and jaw (includes face,head,neck)	Musculoskeletal system
Q75.9	ICD10AM	Congenital malformation of skull and face bones, unspecified	Other anomalies of skull and jaw (includes face,head,neck)	Musculoskeletal system
Q06.1	ICD10AM	Hypoplasia and dysplasia of spinal cord	Other anomalies of spinal cord and nervous system	Nervous system

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APPENDIX 4: (Continued)

Code	Type	Description	Specific condition group	Major diagnostic category
Q06.2	ICD10AM	Diastematomyelia	Other anomalies of spinal cord and nervous system	Nervous system
Q06.8	ICD10AM	Other specified congenital malformations of spinal cord	Other anomalies of spinal cord and nervous system	Nervous system
Q06.9	ICD10AM	Congenital malformation of spinal cord, unspecified	Other anomalies of spinal cord and nervous system	Nervous system
Q07.89	ICD10AM	Other specified congenital malformations of the nervous system	Other anomalies of spinal cord and nervous system	Nervous system
Q07.9	ICD10AM	Congenital malformation of nervous system, unspecified	Other anomalies of spinal cord and nervous system	Nervous system
Q89.09	ICD10AM	Other specified congenital malformations of spleen	Other anomalies of spleen	Other and unspecified anomalies
Q24.5	ICD10AM	Malformation of coronary vessels	Other anomalies of the cardiovascular system	Cardiovascular system
Q25.8	ICD10AM	Other congenital malformations of great arteries	Other anomalies of the cardiovascular system	Cardiovascular system
Q27.1	ICD10AM	Congenital renal artery stenosis	Other anomalies of the cardiovascular system	Cardiovascular system
Q27.3	ICD10AM	Peripheral arteriovenous malformation	Other anomalies of the cardiovascular system	Cardiovascular system
Q27.8	ICD10AM	Other specified congenital malformations of peripheral vascular system	Other anomalies of the cardiovascular system	Cardiovascular system
Q27.9	ICD10AM	Congenital malformation of peripheral vascular system, unspecified	Other anomalies of the cardiovascular system	Cardiovascular system
Q28.2	ICD10AM	Arteriovenous malformation of cerebral vessels	Other anomalies of the cardiovascular system	Cardiovascular system
Q28.3	ICD10AM	Other and unspecified congenital malformations of cerebral vessels	Other anomalies of the cardiovascular system	Cardiovascular system
Q28.9	ICD10AM	Congenital malformation of circulatory system, unspecified	Other anomalies of the cardiovascular system	Cardiovascular system
Q80.9	ICD10AM	Congenital ichthyosis, unspecified	Other anomalies of the integument	Integumentary system
Q81.8	ICD10AM	Other epidermolysis bullosa	Other anomalies of the integument	Integumentary system
Q81.9	ICD10AM	Epidermolysis bullosa, unspecified	Other anomalies of the integument	Integumentary system
Q82.1	ICD10AM	Xeroderma pigmentosum	Other anomalies of the integument	Integumentary system
Q82.3	ICD10AM	Incontinentia pigmenti	Other anomalies of the integument	Integumentary system
Q83.1	ICD10AM	Accessory breast	Other anomalies of the integument	Integumentary system
Q83.8	ICD10AM	Other congenital malformations of breast	Other anomalies of the integument	Integumentary system
Q83.9	ICD10AM	Congenital malformation of breast, unspecified	Other anomalies of the integument	Integumentary system
Q84.0	ICD10AM	Congenital alopecia	Other anomalies of the integument	Integumentary system
Q84.3	ICD10AM	Anonychia	Other anomalies of the integument	Integumentary system
Q84.81	ICD10AM	Aplasia cutis congenita	Other anomalies of the integument	Integumentary system
Q85.81	ICD10AM	Peutz-Jeghers syndrome	Other anomalies of the integument	Integumentary system
Q85.82	ICD10AM	Sturge-Weber(-Dimitri) syndrome	Other anomalies of the integument	Integumentary system
Q22.8	ICD10AM	Other congenital malformations of tricuspid valve	Other anomalies of tricuspid valve	Cardiovascular system
Q22.9	ICD10AM	Congenital malformation of tricuspid valve, unspecified	Other anomalies of tricuspid valve	Cardiovascular system
Q99.8	ICD10AM	Other specified chromosome abnormalities	Other chromosomal anomalies	Chromosomal anomalies
Q99.9	ICD10AM	Chromosomal abnormality, unspecified	Other chromosomal anomalies	Chromosomal anomalies
Q20.6	ICD10AM	Isomerism of atrial appendages	Other congenital anomalies	Other and unspecified anomalies
Q89.79	ICD10AM	Multiple congenital malformations, not elsewhere classified	Other congenital anomalies	Other and unspecified anomalies
Q89.89	ICD10AM	Other specified congenital malformations	Other congenital anomalies	Other and unspecified anomalies
Q89.9	ICD10AM	Congenital malformation, unspecified	Other congenital anomalies	Other and unspecified anomalies
D82.1	ICD10AM	Di George's syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
Q87.04	ICD10AM	Treacher Collins [-Franceschetti] [-Klein] syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
Q87.09	ICD10AM	Other specified congenital malformation syndromes predominantly affecting facial appearance	Other congenital malformation syndromes	Congenital malformation syndromes
Q87.12	ICD10AM	Cornelia de Lange syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
Q87.14	ICD10AM	Prader-Willi syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
Q87.17	ICD10AM	Smith-Lemli-Opitz syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
Q87.19	ICD10AM	Other specified congenital malformation syndromes predominantly associated with short stature	Other congenital malformation syndromes	Congenital malformation syndromes
Q87.21	ICD10AM	Holt-Oram syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
Q87.22	ICD10AM	Klippel-Trenaunay-Weber syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
Q87.31	ICD10AM	Beckwith-Wiedemann syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
Q87.32	ICD10AM	Sotos syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
Q87.4	ICD10AM	Marfan's syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
Q87.5	ICD10AM	Other congenital malformation syndromes with other skeletal changes	Other congenital malformation syndromes	Congenital malformation syndromes
Q87.82	ICD10AM	Laurence-Moon-Biedl syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
Q87.84	ICD10AM	William's syndrome	Other congenital malformation syndromes	Congenital malformation syndromes
Q87.87	ICD10AM	Velocardiofacial syndrome [VCFS]	Other congenital malformation syndromes	Congenital malformation syndromes
Q87.89	ICD10AM	Other specified congenital malformation syndromes, not elsewhere classified	Other congenital malformation syndromes	Congenital malformation syndromes
Q38.3	ICD10AM	Other and unspecified congenital malformations of tongue	Other gastrointestinal anomalies	Gastrointestinal system
Q38.4	ICD10AM	Congenital malformations of salivary glands and ducts	Other gastrointestinal anomalies	Gastrointestinal system
Q38.6	ICD10AM	Other and unspecified congenital malformations of mouth	Other gastrointestinal anomalies	Gastrointestinal system
Q38.8	ICD10AM	Other and unspecified congenital malformations of pharynx	Other gastrointestinal anomalies	Gastrointestinal system
Q39.3	ICD10AM	Congenital stenosis and stricture of oesophagus	Other gastrointestinal anomalies	Gastrointestinal system
Q40.2	ICD10AM	Other specified congenital malformations of stomach	Other gastrointestinal anomalies	Gastrointestinal system
Q40.3	ICD10AM	Congenital malformation of stomach, unspecified	Other gastrointestinal anomalies	Gastrointestinal system

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APPENDIX 4: (Continued)

Code	Type	Description	Specific condition group	Major diagnostic category
Q43.6	ICD10AM	Congenital fistula of rectum and anus	Other gastrointestinal anomalies	Gastrointestinal system
Q20.8	ICD10AM	Other congenital malformations of cardiac chambers and connections	Other malformations cardiac chambers and connections	Cardiovascular system
Q67.59	ICD10AM	Other specified congenital deformity of spine	Other musculoskeletal anomalies	Musculoskeletal system
Q67.6	ICD10AM	Pectus excavatum	Other musculoskeletal anomalies	Musculoskeletal system
Q67.7	ICD10AM	Pectus carinatum	Other musculoskeletal anomalies	Musculoskeletal system
Q67.8	ICD10AM	Other congenital deformities of chest	Other musculoskeletal anomalies	Musculoskeletal system
Q68.1	ICD10AM	Congenital deformities of hand	Other musculoskeletal anomalies	Musculoskeletal system
Q79.6	ICD10AM	Ehlers-Danlos syndrome	Other musculoskeletal anomalies	Musculoskeletal system
Q79.8	ICD10AM	Other congenital malformations of musculoskeletal system	Other musculoskeletal anomalies	Musculoskeletal system
Q79.9	ICD10AM	Congenital malformation of musculoskeletal system, unspecified	Other musculoskeletal anomalies	Musculoskeletal system
Q62.18	ICD10AM	Atresia and stenosis of other and unspecified site of ureter, unilateral	Other obstructive anomalies of renal pelvis and ureter	Genitourinary system
Q04.09	ICD10AM	Other congenital malformations of corpus callosum	Other reduction deformities of brain	Nervous system
Q04.39	ICD10AM	Other reduction anomalies of brain	Other reduction deformities of brain	Nervous system
Q04.4	ICD10AM	Septo-optic dysplasia	Other reduction deformities of brain	Nervous system
Q62.11	ICD10AM	Atresia and stenosis of ureteropelvic junction, unilateral	Pelvic-ureteric junction obstruction	Genitourinary system
Q62.14	ICD10AM	Atresia and stenosis of ureterovesical junction, bilateral	Pelvic-ureteric junction obstruction	Genitourinary system
Q62.39	ICD10AM	Other congenital obstructive defects of renal pelvis and ureter	Pelvic-ureteric junction obstruction	Genitourinary system
E70.1	ICD10AM	Other hyperphenylalaninaemias	Phenylketonuria	Other and unspecified anomalies
Q87.06	ICD10AM	Pierre Robin sequence	Pierre Robin syndrome	Congenital malformation syndromes
Q69.0	ICD10AM	Accessory finger(s)	Polydactyly	Musculoskeletal system
Q69.1	ICD10AM	Accessory thumb(s)	Polydactyly	Musculoskeletal system
Q69.21	ICD10AM	Accessory hallux [halluces]	Polydactyly	Musculoskeletal system
Q69.29	ICD10AM	Other accessory toe(s)	Polydactyly	Musculoskeletal system
Q69.9	ICD10AM	Polydactyly, unspecified	Polydactyly	Musculoskeletal system
Q74.07	ICD10AM	Bifid digit(s) of upper limb	Polydactyly	Musculoskeletal system
Q64.21	ICD10AM	Congenital posterior urethral valves	Posterior urethral valves	Genitourinary system
Q71.1	ICD10AM	Congenital absence of upper arm and forearm with hand present	Reduction deformities of limbs	Musculoskeletal system
Q71.31	ICD10AM	Congenital absence of finger(s) with remainder of hand intact	Reduction deformities of limbs	Musculoskeletal system
Q71.32	ICD10AM	Congenital absence of thumb with all other digits intact	Reduction deformities of limbs	Musculoskeletal system
Q71.33	ICD10AM	Congenital absence of hand and finger(s)	Reduction deformities of limbs	Musculoskeletal system
Q71.4	ICD10AM	Longitudinal reduction defects of radius	Reduction deformities of limbs	Musculoskeletal system
Q71.5	ICD10AM	Longitudinal reduction defects of ulna	Reduction deformities of limbs	Musculoskeletal system
Q71.8	ICD10AM	Other reduction defects of upper limb(s)	Reduction deformities of limbs	Musculoskeletal system
Q71.9	ICD10AM	Reduction defect of upper limb, unspecified	Reduction deformities of limbs	Musculoskeletal system
Q72.31	ICD10AM	Congenital absence of toe(s) with remainder of foot intact	Reduction deformities of limbs	Musculoskeletal system
Q72.4	ICD10AM	Longitudinal reduction defect of femur	Reduction deformities of limbs	Musculoskeletal system
Q72.6	ICD10AM	Longitudinal reduction defect of fibula	Reduction deformities of limbs	Musculoskeletal system
Q72.7	ICD10AM	Split foot	Reduction deformities of limbs	Musculoskeletal system
Q72.8	ICD10AM	Other reduction defects of lower limb(s)	Reduction deformities of limbs	Musculoskeletal system
Q72.9	ICD10AM	Reduction defect of lower limb, unspecified	Reduction deformities of limbs	Musculoskeletal system
Q73.89	ICD10AM	Other reduction defects of unspecified limb(s)	Reduction deformities of limbs	Musculoskeletal system
Q04.33	ICD10AM	Reduction anomalies of cerebellum	Reduction deformity of cerebellum	Nervous system
Q04.01	ICD10AM	Agenesis of corpus callosum	Reduction deformity of corpus callosum	Nervous system
Q60.0	ICD10AM	Renal agenesis, unilateral	Renal agenesis - dysgenesis	Genitourinary system
Q60.2	ICD10AM	Renal agenesis, unspecified	Renal agenesis - dysgenesis	Genitourinary system
Q60.3	ICD10AM	Renal hypoplasia, unilateral	Renal agenesis - dysgenesis	Genitourinary system
Q60.4	ICD10AM	Renal hypoplasia, bilateral	Renal agenesis - dysgenesis	Genitourinary system
Q60.5	ICD10AM	Renal hypoplasia, unspecified	Renal agenesis - dysgenesis	Genitourinary system
Q60.6	ICD10AM	Potter's syndrome	Renal agenesis - dysgenesis	Genitourinary system
Q97.0	ICD10AM	Karyotype 47,XXX	Sex chromosome anomalies	Chromosomal anomalies
Q97.3	ICD10AM	Female with 46,XY karyotype	Sex chromosome anomalies	Chromosomal anomalies
Q98.0	ICD10AM	Klinefelter's syndrome karyotype 47,XXY	Sex chromosome anomalies	Chromosomal anomalies
Q98.1	ICD10AM	Klinefelter's syndrome, male with more than 2 X chromosomes	Sex chromosome anomalies	Chromosomal anomalies
Q98.4	ICD10AM	Klinefelter's syndrome, unspecified	Sex chromosome anomalies	Chromosomal anomalies
Q99.1	ICD10AM	46,XX true hermaphrodite	Sex chromosome anomalies	Chromosomal anomalies
Q89.30	ICD10AM	Situs inversus, unspecified	Situs Inversus	Situs inversus
Q89.31	ICD10AM	Dextrocardia with situs inversus	Situs Inversus	Situs inversus
Q89.39	ICD10AM	Other specified situs inversus	Situs Inversus	Situs inversus
Q05.11	ICD10AM	Thoracic spina bifida with hydrocephalus, open, aperta, not covered with skin or membrane	Spina Bifida	Nervous system

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APPENDIX 4: (Continued)

Code	Type	Description	Specific condition group	Major diagnostic category
Q05.20	ICD10AM	Lumbar spina bifida with hydrocephalus, unspecified whether lesion is open or closed	Spina Bifida	Nervous system
Q05.22	ICD10AM	Lumbar spina bifida with hydrocephalus, closed, cystica, covered with skin or membrane	Spina Bifida	Nervous system
Q05.40	ICD10AM	Unspecified spina bifida with hydrocephalus, unspecified whether lesion is open or closed	Spina Bifida	Nervous system
Q05.60	ICD10AM	Thoracic spina bifida without hydrocephalus, unspecified whether lesion is open or closed	Spina Bifida	Nervous system
Q05.70	ICD10AM	Lumbar spina bifida without hydrocephalus, unspecified whether lesion is open or closed	Spina Bifida	Nervous system
Q05.71	ICD10AM	Lumbar spina bifida without hydrocephalus, open, aperta, not covered with skin or membrane	Spina Bifida	Nervous system
Q05.72	ICD10AM	Lumbar spina bifida without hydrocephalus, closed, cystica, covered with skin or membrane	Spina Bifida	Nervous system
Q05.80	ICD10AM	Sacral spina bifida without hydrocephalus, unspecified whether lesion is open or closed	Spina Bifida	Nervous system
Q05.81	ICD10AM	Sacral spina bifida without hydrocephalus, open, aperta, not covered with skin or membrane	Spina Bifida	Nervous system
Q05.82	ICD10AM	Sacral spina bifida without hydrocephalus, closed, cystica, covered with skin or membrane	Spina Bifida	Nervous system
Q05.90	ICD10AM	Spina bifida, unspecified, unspecified whether lesion is open or closed	Spina Bifida	Nervous system
Q05.92	ICD10AM	Spina bifida, unspecified, closed, cystica, covered with skin or membrane	Spina Bifida	Nervous system
Q07.0	ICD10AM	Arnold-Chiari syndrome	Spina Bifida	Nervous system
Q23.01	ICD10AM	Congenital stenosis of aortic valve	Stenosis of aortic valve	Cardiovascular system
Q25.6	ICD10AM	Stenosis of pulmonary artery	Stenosis of pulmonary artery	Cardiovascular system
Q22.1	ICD10AM	Congenital pulmonary valve stenosis	Stenosis of pulmonary valve	Cardiovascular system
Q70.0	ICD10AM	Fused fingers	Syndactyly	Musculoskeletal system
Q70.2	ICD10AM	Fused toes	Syndactyly	Musculoskeletal system
Q70.4	ICD10AM	Polysyndactyly	Syndactyly	Musculoskeletal system
Q70.9	ICD10AM	Syndactyly, unspecified	Syndactyly	Musculoskeletal system
Q66.00	ICD10AM	Talipes equinovarus, unspecified	Talipes	Musculoskeletal system
Q66.01	ICD10AM	Structural talipes equinovarus	Talipes	Musculoskeletal system
Q66.1	ICD10AM	Talipes calcaneovarus	Talipes	Musculoskeletal system
Q66.4	ICD10AM	Talipes calcaneovalgus	Talipes	Musculoskeletal system
Q21.3	ICD10AM	Tetralogy of Fallot	Tetralogy of Fallot	Cardiovascular system
Q26.2	ICD10AM	Total anomalous pulmonary venous connection	Total or partial anomalous pulmonary venous return	Cardiovascular system
Q26.3	ICD10AM	Partial anomalous pulmonary venous connection	Total or partial anomalous pulmonary venous return	Cardiovascular system
Q39.21	ICD10AM	Congenital tracheo-oesophageal fistula without atresia	Tracheo-oesophageal fistula (TOF) only	Gastrointestinal system
Q20.1	ICD10AM	Double outlet right ventricle	Transposition of great vessels	Cardiovascular system
Q20.3	ICD10AM	Discordant ventriculoarterial connection	Transposition of great vessels	Cardiovascular system
Q20.5	ICD10AM	Discordant atrioventricular connection	Transposition of great vessels	Cardiovascular system
Q92.8	ICD10AM	Other specified trisomies and partial trisomies of autosomes	Trisomies and partial trisomies - remainder	Chromosomal anomalies
Q92.9	ICD10AM	Trisomy and partial trisomy of autosomes, unspecified	Trisomies and partial trisomies - remainder	Chromosomal anomalies
Q91.5	ICD10AM	Trisomy 13, mosaicism	Trisomy 13	Chromosomal anomalies
Q91.7	ICD10AM	Patau's syndrome, unspecified	Trisomy 13	Chromosomal anomalies
Q91.0	ICD10AM	Trisomy 18, meiotic nondisjunction	Trisomy 18	Chromosomal anomalies
Q91.3	ICD10AM	Edwards' syndrome, unspecified	Trisomy 18	Chromosomal anomalies
Q90.9	ICD10AM	Down's syndrome, unspecified	Trisomy 21	Chromosomal anomalies
Q85.1	ICD10AM	Tuberous sclerosis	Tuberous sclerosis	Integumentary system
Q96.3	ICD10AM	Mosaicism, 45,X/46,XX or XY	Turner syndrome	Chromosomal anomalies
Q96.4	ICD10AM	Mosaicism, 45,X/other cell line(s) with abnormal sex chromosome	Turner syndrome	Chromosomal anomalies
Q96.8	ICD10AM	Other variants of Turner's syndrome	Turner syndrome	Chromosomal anomalies
Q96.9	ICD10AM	Turner's syndrome, unspecified	Turner syndrome	Chromosomal anomalies
Q53.10	ICD10AM	Undescended testicle, unilateral, unspecified site	Undescended testis	Genitourinary system
Q53.12	ICD10AM	Undescended testicle, unilateral, inguinal	Undescended testis	Genitourinary system
Q53.13	ICD10AM	Undescended testicle, unilateral, intra-abdominal	Undescended testis	Genitourinary system
Q53.20	ICD10AM	Undescended testicle, bilateral, unspecified site	Undescended testis	Genitourinary system
Q53.21	ICD10AM	Undescended testicle, bilateral, canalicular	Undescended testis	Genitourinary system
Q53.22	ICD10AM	Undescended testicle, bilateral, inguinal	Undescended testis	Genitourinary system
Q53.23	ICD10AM	Undescended testicle, bilateral, intra-abdominal	Undescended testis	Genitourinary system
Q53.90	ICD10AM	Undescended testicle, unspecified laterality, unspecified site	Undescended testis	Genitourinary system
Q53.92	ICD10AM	Undescended testicle, unspecified laterality, inguinal	Undescended testis	Genitourinary system
Q65.3	ICD10AM	Congenital subluxation of hip, unilateral	Unstable hip	Musculoskeletal system
Q65.4	ICD10AM	Congenital subluxation of hip, bilateral	Unstable hip	Musculoskeletal system
Q65.60	ICD10AM	Unstable hip, unspecified	Unstable hip	Musculoskeletal system
Q65.61	ICD10AM	Unstable hip, unilateral	Unstable hip	Musculoskeletal system
Q65.62	ICD10AM	Unstable hip, bilateral	Unstable hip	Musculoskeletal system
Q62.31	ICD10AM	Ectopic ureterocele	Ureterocoele	Genitourinary system
Q62.32	ICD10AM	Orthotopic ureterocele	Ureterocoele	Genitourinary system

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**APPENDIX 4: (Continued)**

Code	Type	Description	Specific condition group	Major diagnostic category
Q87.27	ICD10AM	VATER association	VATER association	Congenital malformation syndromes
Q21.00	ICD10AM	Ventricular septal defect, unspecified	Ventricular septal defect	Cardiovascular system
Q21.01	ICD10AM	Muscular ventricular septal defect	Ventricular septal defect	Cardiovascular system
Q21.02	ICD10AM	Perimembranous ventricular septal defect	Ventricular septal defect	Cardiovascular system
Q21.09	ICD10AM	Other specified ventricular septal defect	Ventricular septal defect	Cardiovascular system

Source: NSW Register of Congenital Conditions (RoCC) 2009 and NSW Admitted Patient Data Collection (APD) 2009–2010, Centre for Epidemiology and Evidence, NSW Ministry of Health.







