CHAPTER 20

BIRTH DEFECTS REGISTRY

Authority

N.J.S.A. 26:8–40 et seq., specifically 26:8–40.26.

Source and Effective Date

R.2000 d.99, effective February 10, 2000.
See: 31 N.J.R. 2863(a), 32 N.J.R. 802(a).

Chapter Expiration Date

In accordance with N.J.S.A. 52:14B-5.1c, Chapter 20, Birth Defects Registry, expires on August 9, 2005. See: 36 N.J.R. 4357(a).

Chapter Historical Note

Chapter 20, Birth Defects Registry, was adopted as R.1985 d.92, effective March 4, 1985. See: 16 N.J.R. 3118(a), 17 N.J.R. 591(a).

Pursuant to Executive Order No. 66(1978), Chapter 20, Birth Defects Registry, was readopted as R.1990 d.187, effective March 2, 1990. See: 21 N.J.R. 3636(a), 22 N.J.R. 1134(c).

Pursuant to Executive Order No. 66(1978), Chapter 20, Birth Defects Registry, was readopted as R.1995 d.182, effective March 2, 1995. See: 27 N.J.R. 269(a), 27 N.J.R. 1410(b).

Pursuant to Executive Order No. 66(1978), Chapter 20, Birth Defects Registry, was readopted as R.2000 d.99, effective February 10, 2000. See: Source and Effective Date. See, also, section annotations.

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SUBCHAPTER 1. LIVE BIRTHS

8:20–1.1 Definitions

The following words and terms when used in this document shall have the following meanings unless the context clearly indicates otherwise.

"Birth defect" means an abnormality of the body's structure or inherent function which is present at birth, whether such abnormality is manifest at the time of delivery or becomes apparent later in life.

"Infant" means a child from birth to one year of age.

8:20–1.2 Reporting requirements

(a) Any infant who is born to a resident of the State of New Jersey, or who becomes a resident of the State before one year of age, and who is diagnosed as having a birth defect either at birth or any time during the first year of life shall be reported to the State Department of Health and Senior Services, Special Child, Adult and Early Intervention Services Program as follows:

1. The conditions listed as Congenital Anomalies (Diagnostic Codes 740.00 through 759.90) in the most recent revision of the International Classification of Diseases, Clinical Modification, shall, except as specified in (a)1ii below, be reported to Special Child, Adult and Early Intervention Services. In addition, there are several other conditions considered to be defects that are not listed under Diagnostic Codes 740.00 through 759.90 which describe Congenital Anomalies. The birth defects listed in (a)1i below shall also, in every case, be reported to Special Child, Adult and Early Intervention Services. The minor conditions listed in (a)1ii below shall not be reported to Special Child, Adult and Early Intervention Services in every case, but only as required in (a)1iii, iv and v below.

i. Congenital anomalies, including, but not limited to, the following:

(1) Anencephalus and similar anomalies, such as craniorachischis and inencephaly.

(2) Spina bifida with and without mention of hydrocephalus.

(3) Other congenital anomalies of the nervous system, such as: encephalocele; microcephalus; reduction deformities of the brain; congenital hydrocephalus; congenital cerebral palsies, congenital muscular dystrophies; and other anomalies, congenital diseases, lesions and any other deformities of the brain, nervous system or spinal cord.

(4) Congenital anomalies of the eye, such as: anophthalmos; microphthalmos; buphthalmos; congenital cataract and lens anomalies; coloboma and other anomalies of the anterior or posterior segment; congenital anomalies of eyelids, lacrimal system and orbit; and any other anomalies of the eye.

(5) Congenital anomalies of the ear, face and neck, such as: anomalies of the ear causing impairment of hearing; accessory auricle and any other anomalies of the ear; branchial cleft cyst or fistula; preauricular sinus; webbing of the neck; and any other anomalies of face and neck.

(6) Bulbus cordis anomalies and anomalies of cardiac septal closure such as: common truncus; transposition of great vessels; Tetralogy of Fallot; common ventricle; ventricular septal defect; ostium secundum type atrial septal defect; endocardial cushion defects; cor biloculare; and any other defects of septal closure.

(7) Other congenital anomalies of the heart, such as: anomalies of pulmonary valve; congenital tricus-

pid atresia and stenosis; Ebstein's anomaly; congenital stenosis of aortic valve; congenital mitral stenosis of aortic valve; congenital mitral stenosis or insufficiency; hypoplastic left heart syndrome; and any other structural anomalies of the heart.

(8) Other congenital anomalies of circulatory system, such as: patent ductus arteriosus (only in infants larger than 2,500 grams); coarctation of aorta and other anomalies of the aorta, aortic arch or atresia and stenosis of the aorta; anomalies of pulmonary artery; anomalies of great veins, absence or hypoplasia of umbilical artery; other anomalies of peripheral vascular system; or other unspecified anomalies of circulatory system.

(9) Congenital anomalies of respiratory system, such as: choanal atresia; other anomalies of nose; webbing of larynx; other anomalies of larynx, trachea and bronchus; congenital cystic lung; agenesis, hypoplasia and dysplasia of lung; other anomalies of the lung; and other unspecified anomalies of respiratory system.

(10) Cleft palate and cleft lip.

(11) Other congenital anomalies of upper alimentary tract, such as: anomalies of the tongue; anomalies of mouth and pharynx; tracheoesophageal fistula, esophageal atresia, and stenosis and other anomalies of esophagus; congenital hypertrophic pyloric stenosis, congenital hiatal hernia; other anomalies of stomach; and other unspecified anomalies of upper alimentary tract.

(12) Other congenital anomalies of digestive system, such as: Meckel's diverticulum; atresia and stenosis of small intestine, large intestine, rectum and anal canal; Hirschsprung's disease and other congenital functional disorders of colon; anomalies of intestinal fixation; other anomalies of intestine, gall bladder, bile ducts, liver and pancreas; disorders of tooth formation, development and eruption, dentofacial anomalies, and other unspecified anomalies of the digestive system.

(13) Congenital anomalies of genital organs, such as: anomalies of ovaries, fallopian tubes and broad ligaments; doubling of uterus and other anomalies of uterus; anomalies of cervix, vagina and external female genitalia; undescended testicle; hypospadias and congenital chordee; indeterminate sex and pseudohermaphroditism; and other unspecified anomalies of the genital system.

(14) Congenital anomalies of urinary system, such as: renal agenesis and dysgenesis; cystic kidney disease; obstructive defects of renal pelvis and ureter; other anomalies of kidney and ureter; exstrophy of urinary bladder; atresia and stenosis of urethra and bladder neck; anomalies of urachus; other anomalies of bladder and urethra; and other unspecified anomalies of the urinary system.

(15) Certain congenital musculoskeletal deformities, such as: of skull, face and jaw; of sternocleidomastoid muscle; of spine; congenital dislocation of hip; congenital genu recurvatum and bowing of long bones of leg; varus and valgus deformities of feet; other congenital deformities of feet such as talipes cavus, calcaneus or equinus; and other specified nonteratogenic anomalies such as pectus excavatum, pectus carinatum; club hand; congenital deformity of chest wall; dislocation of elbow; generalized flexion contractures of lower limbs; spade-like hand.

(16) Other congenital anomalies of limbs, such as: syndactyly; reduction deformities of upper limb; reduction deformities of lower limb; other anomalies of upper limb, including shoulder girdle; and other anomalies of lower limb, including pelvic girdle.

(17) Other congenital musculoskeletal anomalies, such as: anomalies of skull and facial bones; anomalies of spine; cervical rib; other anomalies of ribs and sternum; chondrodystrophy; osteodystrophies; anomalies of diaphragm; anomalies of abdominal wall such as prune belly syndrome; other specified anomalies of muscle, tendon, fascia and connective tissue; and other unspecified anomalies of musculoskeletal system.

(18) Congenital anomalies of the integument, significant anomalies of skin, subcutaneous tissue, hair, nails and breast, such as birthmarks or nevi measuring four inches or greater in size, multiple skin tags (more than five in number).

(19) Chromosomal anomalies, such as: Down's syndrome; Patau's syndrome; Edwards' syndrome; autosomal deletion syndromes and other conditions due to autosomal anomalies; gonadal dysgenesis; Klinefelter's syndrome; and other conditions due to sex chromosome anomalies or anomalies of unspecified chromosome.

(20) Other and unspecified congenital anomalies, such as: anomalies of spleen, situs inversus; conjoined twins; tuberous sclerosis; other hamartoes; multiple congenital anomalies; and other congenital anomalies including congenital malformation syndromes affecting multiple organ systems including Laurence–Moon–Biedl syndrome, Marfan's syndrome and Prader–Willi syndrome.

(21) Certain endocrine, nutritional and metabolic diseases and immunity disorders, includes congenital hypothyroidism; congenital hypoparathyroidism; hypopituitarism; diencephalic syndrome; adrenogenital syndrome; testicular feminization syndrome; phenylketonuria; albinism; maple syrup urine disease; argininosuccinic aciduria; glycogen storage diseases; cystic fibrosis; alpha–1 antitrypsin deficiency; DiGeorge's syndrome; congenital deficiencies of humoral immunity; cell-mediated immunity; combined immunity deficiencies; and other specified and unspecified disorders of the immune mechanisms.

(22) Certain diseases of the blood and blood forming organs, includes hemolytic diseases of the newborn; G–6PD deficiency; hemophilia (all types); Von Willebrand's disease; and sickle-cell anemia or other hemoglobin-apathies.

(23) Certain diseases of the nervous system and sense organs, includes hereditary and degenerative diseases of the central nervous system such as Tay Sachs disease and familial degenerative CNS diseases; Werdnig–Hoffmann disease; cerebral palsy; Moebius syndrome; hereditary retinal dystrophies, and chorioretinitis.

(24) Certain diseases of the circulatory system, includes endocardial fibroelastosis; congenital Wolfe–Parkinson–White syndrome; and Budd–Chiari syndrome.

(25) Certain diseases of the digestive system, includes abnormalities of jaw size, micrognathiae and macrognathia; congenital inguinal hernia with gangrene (only in females), congenital, inguinal hernia with obstruction with no mention of gangrene (only in females), congenital, inguinal hernia without obstruction with no mention of gangrene (only in females), umbilical hernia (only if not covered by skin), epigastric hernia.

(26) Certain complications of pregnancy childbirth, and the puerperium, includes amniotic bands, amniotic cyst.

(27) Certain diseases of the skin and subcutaneous tissue, pilonidal sinus.

(28) Certain conditions originating in the perinatal period, includes fetal alcohol syndrome, probable fetal alcohol syndrome (includes facies), fetal alcohol effects, fetal hydantoin (Dilantin) syndrome, bronchopulmonary dysplasia, unspecified TORCH infection and certain congenital infections including congenital syphilis, congenital rubella, cytomegalovirus, toxoplasmosis, hepatitis, herpes simplex.

(29) Neoplasms, includes lipomas of skin and subcutaneous tissue of face and other skin and subcutaneous tissue, intrathoracic and intra-abdominal organs, spermatic cord, other specified sites, lumbar, sacral, paraspinal, and other unspecified sites; benign neoplasms of skin includes blue nevus, pigmented nevus (include if greater than four inches in diameter), papilloma, dermatofibroma, syringoadenoma, dermoid cyst, hydrocystoma, syringoma; other benign neoplasms of lip, eyelid, ear, external auditory canal, skin and other unspecified parts of face, scalp, skin of neck, skin of trunk, skin of upper limb, skin of lower limb, other specified and unspecified sites including hairy nevus; hemangioma (include if: greater than four inches in diameter, multiple, more than five in number or cavernous hemangioma) of skin and subcutaneous tissue, intracranial, intra-abdominal cystic hygroma; lymphangioma of any site, hemangioma of other and unspecified site; and certain malignant neoplasms including Wilm's tumor, retinoblastoma, other congenital neoplasms including neuroblastoma, medulloblastoma, teratoma, fibrosacrcoma, histiocytosis (malignant), neurofibromatosis.

ii. Minor conditions, as follows:

Accessory auricle
Accessory nipple (supernumerary nipple, or skin tag)
Anal fissure—never a defect
Anal tags
Bat ear
Bell's Palsy
Bent nose, deviation of septum
Big lips
Blue sclera (babies <2500 grams)
Brachial palsy
Breast Hypertrophy—never a defect
Cafe-au-lait spots (register if five or more)
Caput succedaneum
Cardiac murmur [1]
Cauliflower ear
CNS hemorrhage
Cephalhematoma—never a defect
Cervical rib
Chalasia (gastroesophageal reflux)—never a defect
Clinodactyly (incurving of fifth finger)
Congenital hydrocele
Conjunctivitis—never a defect
Cryptorchidism (undescended testicle) [2]
Darwin's tubercle
Diastasis recti—never a defect
Downward eyeslant (antimongoloid)
Ear tags, preauricular tags
Elfin ear
Epicanthal folds
Epulis—never a defect
Erb's palsy
Erythema toxicum
Esotropia
Exotropia
Facial palsy
Flammeus nevus or port wine stain (<four inches in diameter)
Flat bridge or nose
Fontanel (large or small)
Fractured clavicle
Fused eyelids (not a defect if birth weight is <1001 grams)
Gastroesophageal reflux—never a defect
Gum cysts—includes epulis, ranula, mucocele—never a defect
Hemangioma—<four inches in diameter [3]
Hepatomegaly
Hipclick—without follow-up or therapy—not a defect
Hydrocele
Hydrocephaly; acquired
Hymenal tags
Hypoglycemia, idiopathic
Hypoplastic scrotum
Imperforate hymen
Incurving finger (clinodactyly)
Inguinal hernia in male (Note: do not report in females)

Infant of a diabetic mother; asymptomatic
Intussusception
Lanugo, excessive or persistent
Large fontanel
Laryngomalacia or tracheomalacia—never a defect
Long fingers and/or toes
Lop ear
Low set ears
Macrocheilia (big lips)
Meckel's diverticulum
Meconium peritonitis
Meconium plug
Meconium stained skin or nails—never a defect
Metatarus adductus—never a defect
Metatarus varus
Microcheilia (small lips)
Mongolian spots
Mucocele—never a defect
Nasal lacrimal duct obstruction
Nail defects
Natal teeth
Neonatal acne—never a defect
Nystagmus
Orthopedic positional anomalies [4]
Overlapping toes
Overriding (overlapping) sutures—never a defect
Partial syndactyly second and third toes—web extends <one-third length of second toe
Patent ductus arteriosus (PDA) in infants <2500 grams or resolved prior to or at discharge
Patulous lips (wide lips)
Persistent fetal circulation
Petechiae—never a defect
Phimosis—never a defect
Pilonidal dimple
Pilonidal cyst
Pixie-like ear
Pneumothorax
Pointed ear
Polydactyly (postaxial, type B)—skin tags on hands or feet
Posteriorly rotated ears
Preauricular sinus
Pylorospasm (intermittent)
Ranula—never a defect
Rectal fissure
Redundant foreskin
Rockerbottom feet
Sacral dimple
Sebaceous cysts
Simian crease (transverse palmar crease)
Single umbilical artery
Skin cysts
Small fontanel
Small lips
Splenomegaly
Thymic hypertrophy
Tibial torsion
Tongue-tie
Torsion of spermatic cord
Torsion of testes
Tracheomalacia—never a defect
Umbilical cord atrophy
Umbilical hernias (completely covered by skin)
Undescended testicle [2]
Upturned nose
Upward eyeslant (mongoloid)
Vaginal cysts
Vaginal tags
Webbing of neck
Wide nasal bridge
Widely spaced nipples
Widely spaced first and second toes

iii. If a condition or defect listed in (a)1ii above appears as a single defect, a registration form shall not be completed.

iv. If two or more of the conditions listed in (a)1ii above appear, a registration form shall be completed.

v. If a condition or defect listed in (a)1ii above accompanies a condition or defect listed in either Diagnostic Codes 740.00 through 759.90 in the most recent revision of the International Classification of Diseases, Clinical Modification, or in (a)1i above, a registration form shall be completed.

(b) Any live born infant with a birth defect who has not been previously registered and has expired shall be reported. Such reports shall indicate that the infant has expired.

(c) The administrative officer of every health care facility shall be responsible for establishing the reporting procedures for that facility. The reporting procedures must insure that every infant who is initially diagnosed as having a birth defect shall be reported to the Department. All presumptive, tentative, pending, or rule out diagnoses will be reported at the time of discharge, if the child will be diagnosed at a later time or if test results are pending.

(d) Every physician, dentist, certified nurse midwife, advanced practice nurse, and other health care professionals who diagnose or confirm birth defects shall report to the Department each infant diagnosed as having a birth defect not known to be previously reported.

(e) The director of every clinical laboratory shall report to the Department results of postmortem examination from any infant indicating the existence of a birth defect, not known to be previously reported.

(f) The information to be reported shall be provided upon forms supplied by the State Department of Health and Senior Services:

Special Child, Adult and Early Intervention Services
PO Box 364
Trenton, New Jersey 08625–0364

(g) The reports made pursuant to these rules are to be used only by the Department of Health and Senior Services and other agencies that may be designated by the Commissioner of Health and Senior Services and shall not otherwise be divulged or made public so as to disclose the identity of any person; and such reports shall be included under materials available to public inspection pursuant to P.L. 1963, c.73 (N.J.S.A. 47:1A–1 et seq.).

(h) Cytogenetic laboratories shall report the results of all postnatal chromosomal abnormalities.

(i) When a live infant is registered, the Department shall inform the parent or legal guardian of the registration.

(j) Every health care facility and independent clinical laboratory shall allow access to, or provide necessary information on infants with birth defects and other patients specified by characteristics for research studies related to birth defects conducted by the State Department of Health and Senior Services and which have been approved by the State Commissioner of Health and Senior Services after appropriate review for assuring protection of human subjects by the Department's Institutional Review Board. This shall include patients who came under the care of the health facility prior to March 4, 1985.

(k) Any agency designated by the Commissioner to receive reports pursuant to this chapter shall provide to Special Child, Adult and Early Intervention Services any updated diagnostic and/or demographic information.

Amended by R.1987 d.361, effective September 8, 1987.
See: 19 N.J.R. 909(b), 19 N.J.R. 1642(b).

Subsection (a) added a list of congenital anomalies and other conditions which also constituted reportable birth defects.
Amended by R.1990 d.187, effective April 2, 1990.
See: 21 N.J.R. 3636(a), 22 N.J.R. 1134(c).

Reporting requirements for certain conditions specified further; reporting requirements for sickle-cell anemia and other hemoglobinopathies added; all presumptive, tentative, pending and rule out diagnoses to be reported at discharge; cytogenic laboratories to report postnatal chromosomal abnormality test results to the Department.
Amended by R.1991 d.414, effective August 5, 1991.
See: 23 N.J.R. 820(a), 23 N.J.R. 2335(a).

In (a)1, added ii. through v.
Amended by R.1992 d.184, effective April 20, 1992.
See: 24 N.J.R. 171(a), 24 N.J.R. 1494(b).

Minor conditions added at (a)1ii.
Amended by R.2000 d.99, effective March 6, 2000.
See: 31 N.J.R. 2863(a), 32 N.J.R. 802(a).

In (a) and (f), substituted references to Special Child, Adult and Early Intervention Services for references to Special Child Health Services; rewrote (d); in (j), inserted a reference to the Department's Institutional Review Board at the end of the first sentence, and substituted a reference to March 4, 1985 for a reference to the effective date of the regulations at the end of the last sentence; and added (k).

[1] Do not register innocent or functional murmurs: register only if there is a definitive cardiac anomaly or register as a rule out condition if the cause of murmur is not identified at the time of discharge.

[2] Register only if there is clinical evidence of congenital absence.

[3] Register cavernous hemangiomas and multiples of five or more.

[4] Do not register if defect can be corrected passively and does not require casting or bracing.