

MeSH Tree Structures - 2008

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital, Hereditary, and Neonatal Diseases and Abnormalities	C16		
Congenital Abnormalities	C16.131		
Abnormalities, Drug-Induced	C16.131.42		
Abnormalities, Multiple	C16.131.77		
Alagille Syndrome	C16.131.77.65	C6.130.	C6.552.
		C14.240.	C16.131.
		C16.320.	
Angelman Syndrome	C16.131.77.95	C10.228.	C16.131.
		C16.320.	
Bardet-Biedl Syndrome	C16.131.77.112	C10.228.	
Basal Cell Nevus Syndrome	C16.131.77.130	C4.182.	C4.557.
		C4.557.	C4.700.
		C5.116.	C5.500.
		C7.320.	C16.320.
Beckwith-Wiedemann Syndrome	C16.131.77.133	C16.131.	C16.320.
Bloom Syndrome	C16.131.77.137	C18.452.	
Branchio-Oto-Renal Syndrome	C16.131.77.208	C16.131.	C16.320.
Cockayne Syndrome	C16.131.77.250	C5.116.	C10.574.
		C16.320.	C16.320.
		C18.452.	
Cri-du-Chat Syndrome	C16.131.77.262	C10.597.	C16.131.
		C16.320.	
De Lange Syndrome	C16.131.77.272	C10.597.	C16.131.
		C16.320.	
Deaf-Blind Disorders	C16.131.77.299	C9.218.	C10.597.
		C10.597.	C11.966.
		C23.888.	C23.888.
Usher Syndromes	C16.131.77.299.500	C9.218.	C9.218.
		C10.597.	C10.597.
		C10.597.	C11.270.
		C11.768.	C11.966.
		C16.320.	C23.888.
Wolfram Syndrome	C16.131.77.299.750	C9.218.	C10.292.
		C10.574.	C10.597.
		C10.597.	C11.270.
		C11.640.	C11.966.
		C12.777.	C13.351.
		C16.131.	C16.320.
		C16.320.	C18.452.
		C19.246.	C19.700.
Down Syndrome	C16.131.77.327	C10.597.	C16.131.
		C16.320.	
Ectodermal Dysplasia	C16.131.77.350	C16.131.	C16.320.
Ectodermal Dysplasia 1, Anhidrotic	C16.131.77.350.198	C17.800.	C17.800.
		C16.131.	C16.320.
		C16.320.	C17.800.
		C17.800.	
Ectodermal Dysplasia 3, Anhidrotic	C16.131.77.350.298	C16.131.	C16.320.
		C17.800.	C17.800.
Ectodermal Dysplasia, Hypohidrotic, Autosomal Recessive	C16.131.77.350.348	C16.131.	C16.320.
		C17.800.	C17.800.
Ellis-Van Creveld Syndrome	C16.131.77.350.398	C5.116.	C16.131.
		C16.320.	C17.800.
		C17.800.	
Focal Dermal Hypoplasia	C16.131.77.350.424	C5.116.	C16.131.
		C16.320.	C16.320.
		C17.800.	C17.800.
Neurocutaneous Syndromes	C16.131.77.350.712	C10.562	C16.131.
		C16.320.	C17.800.
		C17.800.	

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Congenital Abnormalities
Abnormalities, Multiple
Gardner Syndrome

Gardner Syndrome	C16.131.77.393	C4.557. C4.588. C6.301. C6.405. C6.405.	C4.588. C4.700. C6.405. C6.405. C16.320.
Holoprosencephaly	C16.131.77.410	C5.660. C16.131. C16.131.	C10.500. C16.131. C16.320.
Incontinentia Pigmenti	C16.131.77.445	C16.131. C17.800. C17.800.	C16.320. C17.800. C17.800.
Laurence-Moon Syndrome	C16.131.77.509	C10.228.	
LEOPARD Syndrome	C16.131.77.525	C5.660. C14.280. C16.131. C17.800.	C14.240. C14.280. C16.131.
Marfan Syndrome	C16.131.77.550	C5.116. C14.280. C16.320.	C14.240. C16.131. C17.300.
Mobius Syndrome	C16.131.77.578	C10.292.	C16.614.
Nail-Patella Syndrome	C16.131.77.606	C5.550. C17.800.	C16.320.
Nevus Sebaceous of Jadassohn	C16.131.77.633	C4.557.	C10.562.
Oculocerebrorenal Syndrome	C16.131.77.661	C10.228. C13.351. C16.320. C16.320. C18.452. C18.452.	C12.777. C16.320. C16.320. C18.452. C18.452.
Orofaciodigital Syndromes	C16.131.77.677	C5.116. C16.131. C16.320.	C5.660. C16.131.
POEMS Syndrome	C16.131.77.703	C10.668. C20.683.	C15.378.
Prader-Willi Syndrome	C16.131.77.730	C10.597. C16.320.	C16.131. C18.654.
Proteus Syndrome	C16.131.77.740	C4.445. C5.116. C16.131.	C4.651. C5.660.
Prune Belly Syndrome	C16.131.77.745		
Rubella Syndrome, Congenital	C16.131.77.790	C2.782.	
Rubinstein-Taybi Syndrome	C16.131.77.804	C5.116. C10.597. C16.131.	C5.660. C16.131. C16.320.
Short Rib-Polydactyly Syndrome	C16.131.77.850	C5.116. C16.131.	C5.660.
Smith-Lemli-Opitz Syndrome	C16.131.77.860	C16.320. C18.452. C18.452.	C16.320. C18.452.
Trichothiodystrophy Syndromes	C16.131.77.899	C16.131. C17.800.	C16.320. C17.800.
Waardenburg's Syndrome	C16.131.77.938		
Wolf-Hirschhorn Syndrome	C16.131.77.944	C16.131.	C16.320.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

**Congenital Abnormalities
Abnormalities, Multiple
Wolfram Syndrome**

Wolfram Syndrome

C16.131.77.951

C9.218. C10.292.
C10.574. C10.597.
C10.597. C11.270.
C11.640. C11.966.
C12.777. C13.351.
C16.131. C16.320.
C16.320. C18.452.

Zellweger Syndrome

C16.131.77.970

C19.246. C19.700.
C6.552. C10.228.
C12.777. C13.351.
C16.320. C16.320.
C18.452. C18.452.
C18.452.

**Abnormalities, Radiation-Induced
Cardiovascular Abnormalities
Heart Defects, Congenital
Alagille Syndrome**

C16.131.80

C21.866. G3.850.

C16.131.240

C14.240

C16.131.240.400

C14.240. C14.280.

C16.131.240.400.44

C6.130. C6.552.
C14.240. C16.131.
C16.320.

Aortic Coarctation

C16.131.240.400.90

C14.240. C14.280.

Arrhythmogenic Right Ventricular Dysplasia

C16.131.240.400.145

C14.240. C14.280.
C14.280.

Cor Triatriatum

C16.131.240.400.200

C14.240. C14.280.

Coronary Vessel Anomalies

C16.131.240.400.210

C14.240. C14.280.

Myocardial Bridging

C16.131.240.400.210.500

C14.240. C14.280.

Crisscross Heart

C16.131.240.400.220

C14.240. C14.280.

Dextrocardia

C16.131.240.400.280

C14.240. C14.280.

Kartagener Syndrome

C16.131.240.400.280.500

C16.131.
C8.127. C8.200.
C8.695. C9.150.
C14.240. C14.280.
C16.131. C16.131.
C16.320.

Ductus Arteriosus, Patent

C16.131.240.400.340

C14.240. C14.280.

Ebstein Anomaly

C16.131.240.400.395

C14.240. C14.280.

Ectopia Cordis

C16.131.240.400.422

C14.240.

Eisenmenger Complex

C16.131.240.400.450

C14.240. C14.280.

Heart Septal Defects

C16.131.240.400.560

C14.240. C14.280.

Aortopulmonary Septal Defect

C16.131.240.400.560.98

C14.240. C14.280.

Truncus Arteriosus, Persistent

C16.131.240.400.560.98.500

C14.240. C14.280.

Endocardial Cushion Defects

C16.131.240.400.560.350

C14.240. C14.280.

Heart Septal Defects, Atrial

C16.131.240.400.560.375

C14.240. C14.280.

Foramen Ovale, Patent

C16.131.240.400.560.375.258

C14.240. C14.280.

Lutembacher Syndrome

C16.131.240.400.560.375.518

C14.240. C14.280.

Heart Septal Defects, Ventricular

C16.131.240.400.560.540

C14.240. C14.280.

Hypoplastic Left Heart Syndrome

C16.131.240.400.625

C14.240. C14.280.

LEOPARD Syndrome

C16.131.240.400.685

C5.660. C14.240.
C14.280. C14.280.
C16.131. C16.131.
C17.800.

Levocardia

C16.131.240.400.701

C14.240. C14.280.

Long QT Syndrome

C16.131.240.400.715

C16.131.

Andersen Syndrome

C16.131.240.400.715.70

C14.280. C23.550.

Jervell-Lange Nielsen Syndrome

C16.131.240.400.715.440

C14.280. C23.550.

Romano-Ward Syndrome

C16.131.240.400.715.720

C14.280. C23.550.

Marfan Syndrome

C16.131.240.400.720

C5.116. C14.240.
C14.280. C16.131.

Tetralogy of Fallot

C16.131.240.400.849

C16.320. C17.300.
C14.240. C14.280.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Cardiovascular Abnormalities

Heart Defects, Congenital

Transposition of Great Vessels

Transposition of Great Vessels

Double Outlet Right Ventricle

Tricuspid Atresia

Trilogy of Fallot

Wolff-Parkinson-White Syndrome

Vascular Malformations

Arterio-Arterial Fistula

Arteriovenous Malformations

Arteriovenous Fistula

Intracranial Arteriovenous Malformations

Vein of Galen Malformations

Central Nervous System Vascular Malformations

Intracranial Arteriovenous Malformations

Vein of Galen Malformations

Pulmonary Atresia

Scimitar Syndrome

Telangiectasia, Hereditary Hemorrhagic

Chromosome Disorders

Angelman Syndrome

Beckwith-Wiedemann Syndrome

Branchio-Oto-Renal Syndrome

Cri-du-Chat Syndrome

De Lange Syndrome

Down Syndrome

Holoprosencephaly

Jacobsen Distal 11q Deletion Syndrome

Prader-Willi Syndrome

Rubinstein-Taybi Syndrome

Sex Chromosome Disorders

Fragile X Syndrome

	C16.131.240.400.915	C14.240.	C14.280.
	C16.131.240.400.915.300	C14.240.	C14.280.
	C16.131.240.400.920	C14.240.	C14.280.
		C14.280.	
	C16.131.240.400.960	C14.240.	C14.280.
	C16.131.240.400.980	C14.280.	
	C16.131.240.850	C14.240.	
	C16.131.240.850.500	C14.240.	C14.907.
		C23.300.	
	C16.131.240.850.750	C14.240.	C14.907.
	C16.131.240.850.750.125	C14.240.	C14.240.
		C14.907.	C14.907.
		C23.300.	
	C16.131.240.850.750.295	C10.228.	C10.500.
		C14.240.	C14.240.
		C14.907.	C14.907.
		C16.131.	C16.131.
	C16.131.240.850.750.295.500	C10.228.	C10.500.
		C14.240.	C14.240.
		C14.907.	C14.907.
		C16.131.	C16.131.
	C16.131.240.850.875	C10.500.	C14.240.
		C16.131.	
	C16.131.240.850.875.500	C10.228.	C10.500.
		C14.240.	C14.240.
		C14.907.	C14.907.
		C16.131.	C16.131.
	C16.131.240.850.875.500.500	C10.228.	C10.500.
		C14.240.	C14.240.
		C14.907.	C14.907.
		C16.131.	C16.131.
	C16.131.240.850.906	C14.240.	C14.280.
	C16.131.240.850.937	C8.381.	C8.695.
		C14.240.	C14.907.
		C16.131.	
	C16.131.240.850.968	C14.907.	C14.907.
		C15.378.	
	C16.131.260	C16.320.	
	C16.131.260.40	C10.228.	C16.131.
		C16.320.	
	C16.131.260.80	C16.131.	C16.320.
	C16.131.260.90	C16.131.	C16.320.
	C16.131.260.190	C10.597.	C16.131.
		C16.320.	
	C16.131.260.210	C10.597.	C16.131.
		C16.320.	
	C16.131.260.260	C10.597.	C16.131.
		C16.320.	
	C16.131.260.380	C5.660.	C10.500.
		C16.131.	C16.131.
		C16.131.	C16.320.
	C16.131.260.440	C15.378.	C16.320.
	C16.131.260.700	C10.597.	C16.131.
		C16.320.	C18.654.
	C16.131.260.790	C5.116.	C5.660.
		C10.597.	C16.131.
		C16.131.	C16.320.
	C16.131.260.800	C16.320.	
	C16.131.260.800.300	C10.597.	C16.320.
		C16.320.	C16.320.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Chromosome Disorders

Sex Chromosome Disorders

Gonadal Dysgenesis, 46,XY

Gonadal Dysgenesis, 46,XY

C16.131.260.800.340

C12.706. C13.351.
C16.131. C16.320.
C19.391.

Gonadal Dysgenesis, Mixed

C16.131.260.800.345

C12.706. C13.351.
C16.131. C16.320.
C19.391.

Klinefelter Syndrome

C16.131.260.800.490

C12.706. C16.131.
C16.320. C19.391.
C19.391.

Orofaciodigital Syndromes

C16.131.260.800.670

C5.116. C5.660.
C16.131. C16.131.
C16.320.

Turner Syndrome

C16.131.260.800.870

C12.706. C13.351.
C16.131. C16.320.
C19.391.

WAGR Syndrome

C16.131.260.940

C4.557. C4.588.
C4.700. C10.597.
C11.250. C11.270.
C11.941. C12.706.
C12.758. C12.777.
C13.351. C13.351.
C13.351. C16.131.
C16.131. C16.320.
C16.320. C16.320.

Williams Syndrome

C16.131.260.970

C10.597. C14.280.
C16.320.

Wolf-Hirschhorn Syndrome

C16.131.260.985

C16.131. C16.320.

DiGeorge Syndrome

C16.131.300

C19.642. C20.673.

Digestive System Abnormalities

C16.131.314

C6.198

Anus, Imperforate

C16.131.314.94

C6.198.

Biliary Atresia

C16.131.314.125

C6.130. C6.198.

Choledochal Cyst

C16.131.314.184

C4.182. C6.130.
C6.198.

Caroli Disease

C16.131.314.184.500

C6.130. C6.198.

Diaphragmatic Eventration

C16.131.314.244

C6.198.

Esophageal Atresia

C16.131.314.330

C6.198. C6.405.

Hirschsprung Disease

C16.131.314.439

C6.198. C6.405.

Intestinal Atresia

C16.131.314.466

C6.198. C6.405.

Meckel Diverticulum

C16.131.314.556

A3.556. A3.556.
C6.198. C23.300.

Eye Abnormalities

C16.131.384

C11.250

Aniridia

C16.131.384.79

C11.250. C11.270.
C11.941. C16.320.

WAGR Syndrome

C16.131.384.79.950

C4.557. C4.588.
C4.700. C10.597.
C11.250. C11.270.
C11.941. C12.706.
C12.758. C12.777.
C13.351. C13.351.
C13.351. C16.131.
C16.131. C16.320.
C16.320. C16.320.

Anophthalmos

C16.131.384.159

C11.250.

Blepharophimosis

C16.131.384.190

C11.250. C11.338.

Coloboma

C16.131.384.282

C11.250.

Ectopia Lentis

C16.131.384.405

C11.250. C11.510.

Hydrophthalmos

C16.131.384.480

C11.250. C11.525.
C16.614.

Microphthalmos

C16.131.384.666

C11.250.

Persistent Hyperplastic Primary Vitreous

C16.131.384.725

C11.250.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Eye Abnormalities

Retinal Dysplasia

Retinal Dysplasia	C16.131.384.784	C11.250. C11.768.	C11.270. C16.320.
Lymphatic Abnormalities	C16.131.482	C15.604.	
Lymphangiectasis, Intestinal	C16.131.482.500	C15.604.	C15.604.
Monsters	C16.131.581		
Anencephaly	C16.131.581.197	C10.500.	C16.131.
Twins, Conjoined	C16.131.581.806		
Musculoskeletal Abnormalities	C16.131.621	C5.660	
Arthrogryposis	C16.131.621.77	C5.550. C5.660.	C5.651.
Craniofacial Abnormalities	C16.131.621.207	C5.660.	
Cleidocranial Dysplasia	C16.131.621.207.207	C5.116.	C5.660.
Craniofacial Dysostosis	C16.131.621.207.231	C5.116.	C5.660.
Hallermann's Syndrome	C16.131.621.207.231.427	C5.116.	C5.660.
Hypertelorism	C16.131.621.207.231.480	C5.116.	C5.660.
Mandibulofacial Dysostosis	C16.131.621.207.231.576	C5.116.	C5.660.
Goldenhar Syndrome	C16.131.621.207.231.576.410	C5.116.	C5.660.
Craniosynostoses	C16.131.621.207.240	C5.116. C5.660.	C5.660. C16.131.
Acrocephalosyndactylia	C16.131.621.207.240.100	C5.116. C5.660. C5.660.	C5.116. C5.660. C5.660.
		C16.131.	C16.131.
Holoprosencephaly	C16.131.621.207.410	C5.660. C16.131.	C10.500. C16.131.
		C16.131.	C16.320.
LEOPARD Syndrome	C16.131.621.207.525	C5.660. C14.280.	C14.240. C14.280.
		C16.131.	C16.131.
		C17.800.	
Maxillofacial Abnormalities	C16.131.621.207.540	C5.660.	C7.650.
		C16.131.	
Cherubism	C16.131.621.207.540.170	C5.116.	C5.500.
		C7.320.	C16.320.
Jaw Abnormalities	C16.131.621.207.540.460	C5.500.	C5.660.
		C7.320.	C7.650.
		C16.131.	
Cleft Palate	C16.131.621.207.540.460.185	C5.500. C7.320.	C5.660. C7.465.
		C7.650.	C7.650.
		C16.131.	C16.131.
Micrognathism	C16.131.621.207.540.460.457	C5.500. C7.320.	C5.660. C7.650.
		C16.131.	
Pierre Robin Syndrome	C16.131.621.207.540.460.606	C5.500. C7.320.	C5.660. C7.650.
		C16.131.	
Prognathism	C16.131.621.207.540.460.655	C5.500. C5.660.	C5.500. C7.320.
		C7.320.	C7.650.
		C16.131.	
Retrognathism	C16.131.621.207.540.460.813	C5.500. C5.660.	C5.500. C7.320.
		C7.320.	C7.650.
		C16.131.	
Microcephaly	C16.131.621.207.620	C5.660.	C10.500.
		C16.131.	
Noonan Syndrome	C16.131.621.207.690	C5.660.	C17.300.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Musculoskeletal Abnormalities

Craniofacial Abnormalities

Orofaciodigital Syndromes

Orofaciodigital Syndromes

C16.131.621.207.700

C5.116. C5.660.
C16.131. C16.131.
C16.320.

Plagiocephaly, Nonsynostotic Platybasia

C16.131.621.207.715

C16.131.621.207.720

C5.660. C5.116. C5.116.
C5.660.

Rubinstein-Taybi Syndrome

C16.131.621.207.850

C5.116. C5.660.
C10.597. C16.131.
C16.131. C16.320.

Funnel Chest

Gastroschisis

Hajdu-Cheney Syndrome

C16.131.621.386

C16.131.621.417

C16.131.621.445

C5.116. C5.660.
C5.660. C23.300.
C5.116. C5.116.

Hip Dislocation, Congenital

Klippel-Feil Syndrome

Limb Deformities, Congenital

Arachnodactyly

Ectromelia

Foot Deformities, Congenital

Hand Deformities, Congenital

Lower Extremity Deformities, Congenital

Polydactyly

Short Rib-Polydactyly Syndrome

C16.131.621.449

C16.131.621.551

C16.131.621.585

C16.131.621.585.174

C16.131.621.585.350

C16.131.621.585.380

C16.131.621.585.425

C16.131.621.585.512

C16.131.621.585.600

C16.131.621.585.600.750

C5.550. C5.660.
C5.116. C5.660.

C5.660. C5.660.

C5.660. C5.660.

C5.330. C5.660.
C5.390. C5.660.

C5.660. C5.660.
C5.116. C5.660.

Proteus Syndrome

C16.131.621.585.620

C16.131. C4.445. C4.651.
C5.116. C5.660.

Syndactyly

C16.131.621.585.800

C16.131. C5.116. C5.660.
C5.660. C16.131.

Acrocephalosyndactylyia

C16.131.621.585.800.100

C5.116. C5.116.
C5.660. C5.660.
C5.660. C5.660.

Poland Syndrome

C16.131.621.585.800.756

C16.131. C5.116. C5.660.
C5.660. C16.131.

Thanatophoric Dysplasia

C16.131.621.585.984

C5.116. C5.116.
C5.660. C16.614.

Upper Extremity Deformities, Congenital

Synostosis

Craniosynostoses

Acrocephalosyndactylyia

C16.131.621.585.988

C16.131.621.906

C16.131.621.906.364

C16.131.621.906.364.100

C5.660. C5.660.

C5.116. C5.660.

C5.116. C5.660.
C5.660. C16.131.

C5.116. C5.116.
C5.660. C5.660.

C5.660. C5.660.
C16.131. C16.131.

Syndactyly

C16.131.621.906.819

C16.131. C5.116. C5.660.
C5.660. C16.131.

Acrocephalosyndactylyia

C16.131.621.906.819.100

C5.116. C5.116.
C5.660. C5.660.

C5.660. C5.660.
C16.131. C16.131.

Poland Syndrome

C16.131.621.906.819.756

C5.116. C5.660.
C5.660. C16.131.

C5.116. C5.116.
C5.660. C5.660.

C16.131. C16.131.
C16.131.

C5.116. C5.660.
C5.660. C16.131.

Nervous System Malformations

Central Nervous System Cysts

C16.131.666

C16.131.666.142

C5.116. C5.660.
C10.500

C4.588. C10.500.
C10.551.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Nervous System Malformations

Central Nervous System Cysts

Arachnoid Cysts

Arachnoid Cysts

C16.131.666.142.100

C4.182.

C4.588.

C10.500.

C10.551.

Central Nervous System Vascular Malformations

C16.131.666.190

C10.500.

C14.240.

C16.131.

Central Nervous System Venous Angioma

C16.131.666.190.100

C4.557.

C10.500.

Hemangioma, Cavernous, Central Nervous System

C16.131.666.190.200

C4.557.

C10.500.

C14.240.

C14.907.

C15.378.

Intracranial Arteriovenous Malformations

C16.131.666.190.500

C10.228.

C10.500.

C14.240.

C14.240.

C14.907.

C14.907.

C16.131.

C16.131.

Vein of Galen Malformations

C16.131.666.190.500.500

C10.228.

C10.500.

C14.240.

C14.240.

C14.907.

C14.907.

C16.131.

C16.131.

Sinus Pericranii

C16.131.666.190.800

C10.500.

C14.240.

Dandy-Walker Syndrome

C16.131.666.205

C10.228.

C10.228.

C10.228.

C10.500.

C16.614.

Hereditary Motor and Sensory Neuropathies

C16.131.666.300

C10.500.

C10.574.

C10.668.

C16.320.

Charcot-Marie-Tooth Disease

C16.131.666.300.200

C10.500.

C10.574.

C10.668.

C16.320.

Refsum Disease

C16.131.666.300.780

C10.228.

C10.500.

C10.574.

C10.668.

C16.320.

C16.320.

C16.320.

C18.452.

C18.452.

C18.452.

Spastic Paraplegia, Hereditary

C16.131.666.300.820

C10.500.

C10.574.

C10.668.

C16.320.

Hereditary Sensory and Autonomic Neuropathies

C16.131.666.310

C10.114.

C10.314.

C10.500.

C10.574.

C10.668.

C10.668.

C16.320.

C20.111.

Dysautonomia, Familial

C16.131.666.310.309

C10.114.

C10.177.

C10.314.

C10.500.

C10.574.

C10.668.

C10.668.

C16.320.

Holoprosencephaly

C16.131.666.410

C5.660.

C10.500.

C16.131.

C16.131.

C16.131.

C16.320.

Hydranencephaly

C16.131.666.450

C10.500.

Malformations of Cortical Development

C16.131.666.507

C10.500.

Lissencephaly

C16.131.666.507.186

C10.500.

Classical Lissencephalies and Subcortical Band Heterotopias

C16.131.666.507.186.230

C10.500.

C10.500.

C16.131.

Cobblestone Lissencephaly

C16.131.666.507.186.249

C10.500.

C10.500.

C16.131.

Microcephaly

C16.131.666.507.500

C5.660.

C10.500.

C16.131.

Neuronal Migration Disorders

C16.131.666.507.812

C10.500.

Classical Lissencephalies and Subcortical Band Heterotopias

C16.131.666.507.812.230

C10.500.

C10.500.

C16.131.

Cobblestone Lissencephaly

C16.131.666.507.812.249

C10.500.

C10.500.

C16.131.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Nervous System Malformations

Malformations of Cortical Development

Neuronal Migration Disorders

Periventricular Nodular Heterotopia

Periventricular Nodular Heterotopia
Tuberous Sclerosis

C16.131.666.507.812.750 C10.500.
 C16.131.666.507.875 C4.445. C10.500.
 C10.562. C10.574.
 C16.320.

Neural Tube Defects

Anencephaly

Arnold-Chiari Malformation

Encephalocele

Meningocele

Meningomyelocele

Spinal Dysraphism

Spina Bifida Cystica

Spina Bifida Occulta

Septo-Optic Dysplasia

Respiratory System Abnormalities

Bronchogenic Cyst

C16.131.666.680 C10.500.
 C16.131.666.680.196 C10.500. C16.131.
 C16.131.666.680.291 C10.500.
 C16.131.666.680.488 C10.500.
 C16.131.666.680.598 C10.500.
 C16.131.666.680.610 C10.500.
 C16.131.666.680.800 C10.500.
 C16.131.666.680.800.730 C10.500.
 C16.131.666.680.800.750 C10.500.
 C16.131.666.845 C10.500.
 C16.131.740 C8.695
 C16.131.740.195 C4.182. C8.127.
 C8.695.

Bronchopulmonary Sequestration

Choanal Atresia

C16.131.740.214 C8.695.
 C16.131.740.271 C8.460. C8.695.
 C9.603.

Cystic Adenomatoid Malformation of Lung, Congenital
Kartagener Syndrome

C16.131.740.290 C8.381. C8.695.
 C16.131.740.501 C8.127. C8.200.
 C8.695. C9.150.
 C14.240. C14.280.
 C16.131. C16.131.
 C16.320.

Scimitar Syndrome

C16.131.740.815 C8.381. C8.695.
 C14.240. C14.907.
 C16.131.
 C8.127. C8.695.
 C8.907.

Tracheobronchomegaly

C16.131.740.830 C8.127. C8.695.
 C8.907.

Situs Inversus

Dextrocardia

C16.131.810
 C16.131.810.250 C14.240. C14.280.
 C16.131.

Kartagener Syndrome

C16.131.810.250.500 C8.127. C8.200.
 C8.695. C9.150.
 C14.240. C14.280.
 C16.131. C16.131.
 C16.320.

Levocardia

C16.131.810.700 C14.240. C14.280.
 C16.131.

Skin Abnormalities

Acrodermatitis

Dyskeratosis Congenita

C16.131.831 C17.800.
 C16.131.831.66 C17.800. C17.800.
 C16.131.831.150 C16.320. C16.320.
 C17.800. C17.800.

Ectodermal Dysplasia

C16.131.831.350 C16.131. C16.320.
 C17.800. C17.800.

Ectodermal Dysplasia 1, Anhidrotic

C16.131.831.350.198 C16.131. C16.320.
 C16.320. C17.800.
 C17.800.

Ectodermal Dysplasia 3, Anhidrotic

C16.131.831.350.298 C16.131. C16.320.
 C17.800. C17.800.

Ectodermal Dysplasia, Hypohidrotic, Autosomal Recessive

C16.131.831.350.348 C16.131. C16.320.
 C17.800. C17.800.

Ellis-Van Creveld Syndrome

C16.131.831.350.398 C5.116. C16.131.
 C16.320. C17.800.
 C17.800.

Focal Dermal Hypoplasia

C16.131.831.350.424 C5.116. C16.131.
 C16.320. C16.320.
 C17.800. C17.800.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Skin Abnormalities

Ectodermal Dysplasia

Neurocutaneous Syndromes

Neurocutaneous Syndromes	C16.131.831.350.712	C10.562 C16.320. C17.800.	C16.131. C17.800.
Pachyonychia Congenita	C16.131.831.350.856	C16.320. C17.800.	C17.800. C17.800.
Ehlers-Danlos Syndrome	C16.131.831.428	C14.907. C16.320. C17.800.	C15.378. C17.300. C17.800.
Epidermolysis Bullosa	C16.131.831.493	C16.320. C17.800.	C17.800. C17.800.
Epidermolysis Bullosa Acquisita	C16.131.831.493.80	C17.800. C17.800.	C17.800. C17.800.
Epidermolysis Bullosa Dystrophica	C16.131.831.493.160	C16.320. C17.800. C17.800.	C17.300. C17.800.
Epidermolysis Bullosa, Junctional	C16.131.831.493.170	C16.320. C17.800.	C17.800. C17.800.
Epidermolysis Bullosa Simplex	C16.131.831.493.180	C16.320. C17.800.	C17.800. C17.800.
Ichthyosis	C16.131.831.512	C16.614. C17.800.	C17.800.
Ichthyosiform Erythroderma, Congenital	C16.131.831.512.400	C16.320. C17.800. C17.800.	C16.614. C17.800.
Hyperkeratosis, Epidermolytic	C16.131.831.512.400.375	C16.320. C17.800. C17.800.	C16.614. C17.800.
Ichthyosis, Lamellar	C16.131.831.512.400.410	C16.320. C17.800. C17.800.	C16.614. C17.800.
Ichthyosis Bullosa of Siemens	C16.131.831.512.408	C16.320. C17.800. C17.800.	C16.614. C17.800.
Ichthyosis Vulgaris	C16.131.831.512.410	C16.320. C17.800.	C17.800.
Ichthyosis, X-Linked	C16.131.831.512.420	C16.320. C16.320. C17.800. C17.800.	C16.320. C16.614. C17.800. C18.452.
Sjogren-Larsson Syndrome	C16.131.831.512.723	C16.320. C16.614. C17.800. C18.452.	C16.320. C17.800. C17.800. C18.452.
Incontinentia Pigmenti	C16.131.831.580	C16.131. C17.800. C17.800.	C16.320. C17.800.
Port-Wine Stain	C16.131.831.675	C17.800.	
Pseudoxanthoma Elasticum	C16.131.831.766	C14.907. C16.320. C17.800.	C15.378. C17.300. C17.800.
Rothmund-Thomson Syndrome	C16.131.831.775	C16.320. C17.800. C18.452.	C16.614. C17.800.
Sclerema Neonatorum	C16.131.831.812	C16.614.	C17.800.
Trichothiodystrophy Syndromes	C16.131.831.874	C16.131. C17.800.	C16.320. C17.800.
Xeroderma Pigmentosum	C16.131.831.936	C4.834. C17.800. C17.800. C18.452.	C16.320. C17.800. C17.800.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Stomatognathic System Abnormalities

Stomatognathic System Abnormalities	C16.131.850	C7.650	
Maxillofacial Abnormalities	C16.131.850.500	C5.660.	C7.650.
Jaw Abnormalities		C16.131.	
Cleft Palate	C16.131.850.500.460	C5.500.	C5.660.
Micrognathism		C7.320.	C7.650.
Pierre Robin Syndrome		C16.131.	
Prognathism	C16.131.850.500.460.185	C5.500.	C5.660.
Retrognathism		C7.320.	C7.465.
Cleft Lip		C7.650.	C7.650.
Cleft Palate	C16.131.850.500.460.457	C16.131.	C16.131.
Fibromatosis, Gingival		C5.500.	C5.660.
Macrostomia		C7.320.	C7.650.
Microstomia	C16.131.850.500.460.606	C16.131.	C16.131.
Velopharyngeal Insufficiency		C5.500.	C5.500.
Teeth Abnormalities	C16.131.850.500.460.655	C5.660.	C7.320.
Amelogenesis Imperfecta		C7.320.	C7.650.
Dental Enamel Hypoplasia		C16.131.	
Anodontia	C16.131.850.500.460.813	C5.500.	C5.500.
Dens in Dente		C5.500.	C5.500.
Dentin Dysplasia		C5.660.	C7.320.
Dentinogenesis Imperfecta		C7.320.	C7.650.
Fused Teeth		C16.131.	
Odontodysplasia		C5.500.	C5.500.
Tooth, Supernumerary		C5.660.	C7.320.
Mouth Abnormalities	C16.131.850.525	C7.465.	C7.650.
Cleft Lip		C7.465.	C7.465.
Cleft Palate	C16.131.850.525.164	C7.650.	
Fibromatosis, Gingival		C16.131.	
Macrostomia		C7.465.	C7.465.
Microstomia	C16.131.850.525.304	C7.650.	
Velopharyngeal Insufficiency		C7.650.	
Teeth Abnormalities	C16.131.850.525.480	C7.465.	C7.650.
Amelogenesis Imperfecta		C7.465.	C7.650.
Dental Enamel Hypoplasia	C16.131.850.525.520	C7.465.	C7.550.
Anodontia		C7.650.	C9.775.
Dens in Dente	C16.131.850.525.955	C7.650.	C7.793.
Dentin Dysplasia		C7.650.	C7.793.
Dentinogenesis Imperfecta		C7.650.	C7.793.
Fused Teeth		C7.650.	C7.793.
Odontodysplasia		C7.650.	C7.793.
Tooth, Supernumerary		C7.650.	C7.793.
Thyroid Dysgenesis	C16.131.894	C19.874.	
Lingual Thyroid		C19.874.	
Lingual Goiter	C16.131.894.500	C19.874.	C19.874.
Urogenital Abnormalities	C16.131.939	C12.706	C13.351.
Bladder Exstrophy		C12.706.	C12.777.
Cryptorchidism	C16.131.939.132	C13.351.	C13.351.
Epispadias		C12.294.	C12.706.
Frasier Syndrome		C19.391.	
Hypospadias	C16.131.939.258	C12.706.	C12.777.
		C13.351.	C13.351.
	C16.131.939.374	C12.706.	C13.351.
		C12.706.	C13.351.
	C16.131.939.445	C12.294.	C12.706.
		C13.351.	C13.351.
	C16.131.939.516	C12.294.	C12.706.
		C13.351.	

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Urogenital Abnormalities

Multicystic Dysplastic Kidney

Multicystic Dysplastic Kidney

C16.131.939.629

C12.706.
C13.351.

C12.777.
C13.351.

Nephritis, Hereditary

C16.131.939.742

C12.706.
C13.351.
C17.300.

C12.777.
C13.351.

Sex Differentiation Disorders

C16.131.939.842

C12.706.
C19.391.

C13.351.

Freemartinism

C16.131.939.842.260

C13.351.
C22.196.

C19.391.

Gonadal Dysgenesis

C16.131.939.842.309

C12.706.
C19.391.

C13.351.

Gonadal Dysgenesis, 46,XX

C16.131.939.842.309.193

C12.706.
C19.391.

C13.351.

Gonadal Dysgenesis, 46,XY

C16.131.939.842.309.388

C12.706.
C16.131.

C13.351.
C16.320.

Gonadal Dysgenesis, Mixed

C16.131.939.842.309.391

C19.391.
C12.706.
C16.131.

C13.351.
C16.320.

Turner Syndrome

C16.131.939.842.309.872

C12.706.
C16.131.

C13.351.
C16.320.

Hermaphroditism

C16.131.939.842.316

C12.706.
C19.391.

C13.351.

Hermaphroditism, True

C16.131.939.842.316.313

C12.706.
C19.391.

C13.351.

Pseudohermaphroditism

C16.131.939.842.316.627

C12.706.
C19.391.

C13.351.

Androgen-Insensitivity Syndrome

C16.131.939.842.316.627.109

C12.706.
C16.320.

C13.351.
C19.391.

Denys-Drash Syndrome

C16.131.939.842.316.627.220

C4.557.
C4.700.

C4.588.
C12.706.

C12.758.
C13.351.

C12.777.
C13.351.

Kallmann Syndrome

C16.131.939.842.425

C12.706.
C16.320.

C13.351.
C19.391.

Klinefelter Syndrome

C16.131.939.842.454

C12.706.
C16.320.

C16.131.
C19.391.

WAGR Syndrome

C16.131.939.921

C4.557.
C4.700.

C4.588.
C10.597.

C11.250.
C11.941.

C11.270.
C12.706.

C12.758.
C13.351.

C12.777.
C13.351.

C13.351.
C16.131.

C16.131.
C16.320.

C16.320.
C13.703.

C16.320.

Fetal Diseases

C16.300

C13.703.

Chorioamnionitis

C16.300.30

C13.703.

C13.703.

Erythroblastosis, Fetal

C16.300.60

C13.703.

C15.378.

Hydrops Fetalis

C16.300.60.480

C16.614.

C20.188.

C13.703.

C15.378.

C15.378.

C16.320.

C20.188.

C23.888.

Fetal Alcohol Syndrome

C16.300.80

C13.703.

C21.739.

Fetal Growth Retardation

C16.300.390

C13.703.

C23.550.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Fetal Diseases

Fetal Hypoxia

Fetal Hypoxia	C16.300.420	C13.703.	C23.888.
Fetal Macrosomia	C16.300.570	C13.703.	C13.703.
		C13.703.	C19.246.
		C23.888.	
Meconium Aspiration Syndrome	C16.300.580	C8.381.	C8.618.
		C13.703.	C16.614.
Nuchal Cord	C16.300.790	C13.703.	
Genetic Diseases, Inborn	C16.320		
Adrenal Hyperplasia, Congenital	C16.320.33	C16.320.	C18.452.
		C19.53.	
Alagille Syndrome	C16.320.51	C6.130.	C6.552.
		C14.240.	C16.131.
		C16.131.	
Anemia, Hemolytic, Congenital	C16.320.70	C15.378.	
Anemia, Dyserythropoietic, Congenital	C16.320.70.95	C15.378.	
Anemia, Hemolytic, Congenital Nonspherocytic	C16.320.70.100	C15.378.	
Anemia, Sickle Cell	C16.320.70.150	C15.378.	C15.378.
		C16.320.	
Hemoglobin SC Disease	C16.320.70.150.440	C15.378.	C15.378.
		C16.320.	
Sickle Cell Trait	C16.320.70.150.670	C15.378.	C15.378.
		C16.320.	
Elliptocytosis, Hereditary	C16.320.70.365	C15.378.	
Glucosephosphate Dehydrogenase Deficiency	C16.320.70.480	C15.378.	C16.320.
		C18.452.	
Favism	C16.320.70.480.370	C15.378.	C15.378.
		C21.613.	C21.613.
Hemoglobin C Disease	C16.320.70.490	C15.378.	C15.378.
		C16.320.	
Spherocytosis, Hereditary	C16.320.70.785	C15.378.	
Thalassemia	C16.320.70.875	C15.378.	C15.378.
		C16.320.	
alpha-Thalassemia	C16.320.70.875.100	C15.378.	C15.378.
		C16.320.	
beta-Thalassemia	C16.320.70.875.150	C15.378.	C15.378.
		C16.320.	
Anemia, Hypoplastic, Congenital	C16.320.77	C15.378.	C15.378.
Anemia, Diamond-Blackfan	C16.320.77.90	C15.378.	C15.378.
		C15.378.	
Fanconi Anemia	C16.320.77.280	C15.378.	C15.378.
		C18.452.	
Angioedema, Hereditary	C16.320.78	C14.907.	C17.800.
		C20.543.	
Ataxia Telangiectasia	C16.320.80	C10.228.	C10.562.
		C10.597.	C14.907.
		C18.452.	C20.673.
Blood Coagulation Disorders, Inherited	C16.320.99	C15.378.	
Activated Protein C Resistance	C16.320.99.37	C15.378.	C15.378.
		C15.378.	
Afibrinogenemia	C16.320.99.56	C15.378.	C15.378.
		C15.378.	
Antithrombin III Deficiency	C16.320.99.75	C15.378.	C15.378.
		C15.378.	
Bernard-Soulier Syndrome	C16.320.99.80	C15.378.	C15.378.
		C15.378.	
Factor V Deficiency	C16.320.99.300	C15.378.	C15.378.
		C15.378.	
Factor VII Deficiency	C16.320.99.310	C15.378.	C15.378.
		C15.378.	

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Blood Coagulation Disorders, Inherited

Factor X Deficiency

Factor X Deficiency	C16.320.99.320	C15.378. C15.378.	C15.378.
Factor XI Deficiency	C16.320.99.325	C15.378. C15.378.	C15.378.
Factor XII Deficiency	C16.320.99.330	C15.378. C15.378.	C15.378.
Factor XIII Deficiency	C16.320.99.335	C15.378. C15.378.	C15.378.
Hemophilia A	C16.320.99.500	C15.378. C15.378.	C15.378.
Hemophilia B	C16.320.99.510	C15.378. C15.378.	C15.378.
Hermanski-Pudlak Syndrome	C16.320.99.515	C11.270. C15.378. C15.378. C16.320. C17.800. C18.452.	C15.378. C15.378. C16.320. C17.800.
Hypoprothrombinemias	C16.320.99.550	C15.378. C15.378.	C15.378.
Protein C Deficiency	C16.320.99.690	C15.378. C15.378.	C15.378.
Thrombasthenia	C16.320.99.820	C15.378. C15.378.	C15.378.
von Willebrand Disease	C16.320.99.900	C15.378. C15.378.	C15.378.
Wiskott-Aldrich Syndrome	C16.320.99.970	C15.378. C15.378. C16.320.	C15.378. C15.378. C20.673.
Brugada Syndrome	C16.320.100	C14.280.	
CADASIL	C16.320.129	C10.228. C10.228.	C10.228. C14.907.
Camurati-Engelmann Syndrome	C16.320.144	C5.116.	
Cardiomyopathy, Hypertrophic, Familial	C16.320.160	C14.280.	
Cherubism	C16.320.170	C5.116. C7.320.	C5.500. C16.131.
Chromosome Disorders	C16.320.180	C16.131.	
Angelman Syndrome	C16.320.180.40	C10.228. C16.131.	C16.131.
Beckwith-Wiedemann Syndrome	C16.320.180.80	C16.131.	C16.131.
Branchio-Oto-Renal Syndrome	C16.320.180.90	C16.131.	C16.131.
Cri-du-Chat Syndrome	C16.320.180.190	C10.597. C16.131.	C16.131.
De Lange Syndrome	C16.320.180.210	C10.597. C16.131.	C16.131.
Down Syndrome	C16.320.180.260	C10.597. C16.131.	C16.131.
Holoprosencephaly	C16.320.180.380	C5.660. C16.131. C16.131.	C10.500. C16.131. C16.131.
Jacobsen Distal 11q Deletion Syndrome	C16.320.180.440	C15.378.	C16.131.
Prader-Willi Syndrome	C16.320.180.700	C10.597. C16.131.	C16.131. C18.654.
Rubinstein-Taybi Syndrome	C16.320.180.790	C5.116. C10.597. C16.131.	C5.660. C16.131. C16.131.
Sex Chromosome Disorders	C16.320.180.800	C16.131.	
Fragile X Syndrome	C16.320.180.800.300	C10.597. C16.320.	C16.131. C16.320.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Chromosome Disorders

Sex Chromosome Disorders

Gonadal Dysgenesis, 46,XY

Gonadal Dysgenesis, 46,XY

C16.320.180.800.340

C12.706. C13.351.
C16.131. C16.131.
C19.391.

Gonadal Dysgenesis, Mixed

C16.320.180.800.345

C12.706. C13.351.
C16.131. C16.131.
C19.391.

Klinefelter Syndrome

C16.320.180.800.490

C12.706. C16.131.
C16.131. C19.391.
C19.391.

Orofaciodigital Syndromes

C16.320.180.800.670

C5.116. C5.660.
C16.131. C16.131.
C16.131.

Turner Syndrome

C16.320.180.800.870

C12.706. C13.351.
C16.131. C16.131.
C19.391.

WAGR Syndrome

C16.320.180.940

C4.557. C4.588.
C4.700. C10.597.
C11.250. C11.270.
C11.941. C12.706.
C12.758. C12.777.
C13.351. C13.351.
C13.351. C16.131.
C16.131. C16.131.
C16.320. C16.320.

Williams Syndrome

C16.320.180.970

C10.597. C14.280.
C16.131.

Wolf-Hirschhorn Syndrome

C16.320.180.985

C16.131. C16.131.

Cystic Fibrosis

C16.320.190

C6.689. C8.381.
C16.614.

Dwarfism

C16.320.240

C5.116. C19.297

Achondroplasia

C16.320.240.500

C5.116. C5.116.

Cockayne Syndrome

C16.320.240.562

C5.116. C10.574.
C16.131. C16.320.
C18.452.

Congenital Hypothyroidism

C16.320.240.625

C5.116. C5.116.
C19.297. C19.874.

Laron Syndrome

C16.320.240.750

C5.116. C19.297.

Mulibrey Nanism

C16.320.240.875

C5.116.

Eye Diseases, Hereditary

C16.320.290

C11.270

Albinism

C16.320.290.40

C11.270. C16.320.
C16.320. C17.800.
C17.800. C18.452.

Albinism, Ocular

C16.320.290.40.90

C11.270. C16.320.
C16.320. C17.800.
C17.800. C18.452.

Albinism, Oculocutaneous

C16.320.290.40.100

C11.270. C16.320.
C16.320. C17.800.
C17.800. C18.452.

Hermanski-Pudlak Syndrome

C16.320.290.40.100.400

C11.270. C15.378.
C15.378. C15.378.
C15.378. C16.320.
C16.320. C16.320.

Piebaldism

C16.320.290.40.600

C17.800. C17.800.
C18.452.

Aniridia

C16.320.290.78

C11.250. C11.270.
C11.941. C16.131.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Eye Diseases, Hereditary

Aniridia

WAGR Syndrome

WAGR Syndrome

C16.320.290.78.950

C4.557. C4.588.
C4.700. C10.597.
C11.250. C11.270.
C11.941. C12.706.
C12.758. C12.777.
C13.351. C13.351.
C13.351. C16.131.
C16.131. C16.131.
C16.320. C16.320.
C11.270. C11.941.
C16.320.

Choroideremia

C16.320.290.142

C11.270. C11.270.
C16.320.

Corneal Dystrophies, Hereditary

C16.320.290.162

C11.204. C11.270.

Corneal Dystrophy, Juvenile Epithelial of Meesmann

C16.320.290.162.204

C11.204. C11.270.

Fuchs' Endothelial Dystrophy

C16.320.290.162.410

C11.204. C11.270.

Duane Retraction Syndrome

C16.320.290.235

C10.292. C11.270.
C11.590.

Gyrate Atrophy

C16.320.290.468

C11.270. C11.941.

Optic Atrophies, Hereditary

C16.320.290.564

C10.292. C10.574.
C11.270. C11.640.
C16.320.

Optic Atrophy, Autosomal Dominant

C16.320.290.564.100

C10.292. C10.574.
C11.270. C11.640.
C16.320. C18.452.

Optic Atrophy, Hereditary, Leber

C16.320.290.564.400

C10.292. C10.574.
C11.270. C11.640.
C16.320. C18.452.

Wolfram Syndrome

C16.320.290.564.980

C9.218. C10.292.
C10.574. C10.597.
C10.597. C11.270.
C11.640. C11.966.
C12.777. C13.351.
C16.131. C16.131.
C16.320. C18.452.
C19.246. C19.700.
C11.250. C11.270.
C11.768. C16.131.

Retinal Dysplasia

C16.320.290.660

C11.270. C11.768.
C9.218. C9.218.
C10.597. C10.597.
C10.597. C11.270.
C11.768. C11.966.
C16.131. C23.888.
C20.111.

Retinitis Pigmentosa

C16.320.290.684

Usher Syndromes

C16.320.290.684.500

Familial Mediterranean Fever

C16.320.306

Genetic Diseases, X-Linked

C16.320.322

Androgen-Insensitivity Syndrome

C16.320.322.61

C12.706. C13.351.
C16.131. C19.391.

Choroideremia

C16.320.322.92

C11.270. C11.941.
C16.320.

Dyskeratosis Congenita

C16.320.322.108

C16.131. C16.320.
C17.800. C17.800.

Ectodermal Dysplasia 1, Anhidrotic

C16.320.322.116

C16.131. C16.131.
C16.320. C17.800.
C17.800.

Fabry Disease

C16.320.322.124

C10.228. C16.320.
C16.320. C16.320.
C18.452. C18.452.
C18.452. C18.452.
C18.452.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Genetic Diseases, X-Linked

Focal Dermal Hypoplasia

Focal Dermal Hypoplasia	C16.320.322.186	C5.116. C16.131. C16.320. C17.800. C10.597. C16.320.	C16.131. C16.320. C17.800. C14.280. C18.452.
Glycogen Storage Disease Type IIb	C16.320.322.201	C16.320. C15.378. C15.378. C15.378.	C18.452. C20.673. C15.378. C16.320.
Glycogen Storage Disease Type VIII	C16.320.322.217	C16.320.	C18.452.
Granulomatous Disease, Chronic	C16.320.322.233	C15.378.	C20.673.
Hemophilia B	C16.320.322.235	C15.378. C15.378.	C15.378. C16.320.
Hyper-IgM Immunodeficiency Syndrome, Type 1	C16.320.322.237	C15.378.	C20.673.
Hypophosphatemic Rickets, X-Linked Dominant	C16.320.322.239	C5.116. C13.351. C16.320. C18.452. C18.452. C18.654.	C12.777. C16.320. C18.452. C18.452.
Ichthyosis, X-Linked	C16.320.322.241	C16.131. C16.320. C17.800. C17.800.	C16.320. C16.614. C17.800. C18.452.
Mental Retardation, X-Linked	C16.320.322.500	C10.597.	C16.320.
Adrenoleukodystrophy	C16.320.322.500.124	C10.228. C10.597. C16.320. C18.452. C18.452.	C10.314. C16.320. C16.320. C18.452. C19.53.
Coffin-Lowry Syndrome	C16.320.322.500.249	C10.597.	C16.320.
Fragile X Syndrome	C16.320.322.500.500	C10.597. C16.320.	C16.131. C16.320.
Lesch-Nyhan Syndrome	C16.320.322.500.625	C10.228. C10.597. C16.320. C16.320. C18.452.	C10.574. C16.320. C16.320. C18.452.
Menkes Kinky Hair Syndrome	C16.320.322.500.687	C18.452. C10.228. C10.597. C16.320. C16.320. C18.452.	C18.452. C10.574. C16.320. C16.320. C17.800. C18.452.
Mucopolysaccharidosis II	C16.320.322.500.750	C10.597. C16.320. C17.300. C18.452.	C16.320. C16.320. C18.452.
Pyruvate Dehydrogenase Complex Deficiency Disease	C16.320.322.500.875	C10.228. C16.320. C16.320. C18.452. C18.452.	C10.597. C16.320. C18.452. C18.452.
Rett Syndrome	C16.320.322.500.937	C10.574. C16.320. F3.550.	C10.597. C16.320.
Muscular Dystrophy, Duchenne	C16.320.322.562	C5.651. C16.320.	C10.668.
Muscular Dystrophy, Emery-Dreifuss	C16.320.322.625	C5.651. C16.320.	C10.668.
Oculocerebrorenal Syndrome	C16.320.322.750	C10.228. C13.351. C16.320. C16.320. C18.452. C18.452.	C12.777. C16.131. C16.320. C18.452. C18.452.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Genetic Diseases, X-Linked

Pelizaeus-Merzbacher Disease

Pelizaeus-Merzbacher Disease	C16.320.322.906	C10.314.	
Wiskott-Aldrich Syndrome	C16.320.322.937	C15.378.	C15.378.
		C15.378.	C15.378.
		C16.320.	C20.673.
X-Linked Combined Immunodeficiency Diseases	C16.320.322.968	C16.614.	C20.673.
Genetic Diseases, Y-Linked	C16.320.338		
Hajdu-Cheney Syndrome	C16.320.355	C5.116.	C5.116.
		C16.131.	
Hemoglobinopathies	C16.320.365	C15.378.	
Anemia, Sickle Cell	C16.320.365.155	C15.378.	C15.378.
		C16.320.	
Hemoglobin SC Disease	C16.320.365.155.440	C15.378.	C15.378.
		C16.320.	
Sickle Cell Trait	C16.320.365.155.668	C15.378.	C15.378.
		C16.320.	
Hemoglobin C Disease	C16.320.365.463	C15.378.	C15.378.
		C16.320.	
Thalassemia	C16.320.365.826	C15.378.	C15.378.
		C16.320.	
alpha-Thalassemia	C16.320.365.826.100	C15.378.	C15.378.
		C16.320.	
Hydrops Fetalis	C16.320.365.826.100.350	C13.703.	C15.378.
		C15.378.	C16.300.
		C20.188.	C23.888.
beta-Thalassemia	C16.320.365.826.150	C15.378.	C15.378.
		C16.320.	
Heredodegenerative Disorders, Nervous System	C16.320.400	C10.574.	
Alexander Disease	C16.320.400.24	C10.314.	C10.574.
Amyloid Neuropathies, Familial	C16.320.400.50	C10.574.	C10.668.
		C16.320.	C18.452.
		C18.452.	C18.452.
Canavan Disease	C16.320.400.150	C10.314.	C10.574.
Cockayne Syndrome	C16.320.400.200	C5.116.	C10.574.
		C16.131.	C16.320.
		C18.452.	
Dystonia Musculorum Deformans	C16.320.400.330	C10.228.	C10.228.
		C10.574.	
Gerstmann-Straussler-Scheinker Disease	C16.320.400.350	C10.228.	C10.574.
		C10.574.	
Hepatolenticular Degeneration	C16.320.400.361	C6.552.	C10.228.
		C10.228.	C10.228.
		C10.574.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
Hereditary Central Nervous System Demyelinating Diseases	C16.320.400.367	C10.314.	C10.574.
Pantothenate Kinase-Associated Neurodegeneration	C16.320.400.375	C10.228.	C10.228.
		C10.228.	C10.574.
Hereditary Motor and Sensory Neuropathies	C16.320.400.400	C10.500.	C10.574.
		C10.668.	C16.131.
Charcot-Marie-Tooth Disease	C16.320.400.400.200	C10.500.	C10.574.
		C10.668.	C16.131.
Refsum Disease	C16.320.400.400.780	C10.228.	C10.500.
		C10.574.	C10.668.
		C16.131.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
Spastic Paraplegia, Hereditary	C16.320.400.400.820	C10.500.	C10.574.
		C10.668.	C16.131.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Hereditary Degenerative Disorders, Nervous System
Hereditary Sensory and Autonomic Neuropathies

Hereditary Sensory and Autonomic Neuropathies	C16.320.400.415	C10.114. C10.500. C10.668. C16.131.	C10.314. C10.574. C10.668. C20.111.
Dysautonomia, Familial	C16.320.400.415.309	C10.114. C10.314. C10.574. C10.668. C20.111.	C10.177. C10.500. C10.668. C16.131.
Huntington Disease	C16.320.400.430	C10.228. C10.228. F3.87.	C10.228. C10.574. F3.87.
Lafora Disease	C16.320.400.480	C10.228.	C10.574.
Lesch-Nyhan Syndrome	C16.320.400.500	C10.228. C10.597. C16.320. C16.320. C16.320. C18.452. C18.452.	C10.574. C16.320. C16.320. C18.452. C18.452.
Menkes Kinky Hair Syndrome	C16.320.400.520	C10.228. C10.597. C16.320. C16.320. C16.320. C18.452. C18.452.	C10.574. C16.320. C16.320. C17.800. C18.452.
Mental Retardation, X-Linked Adrenoleukodystrophy	C16.320.400.525 C16.320.400.525.124	C10.597. C10.228. C10.597. C16.320. C16.320. C18.452. C18.452.	C16.320. C10.314. C16.320. C16.320. C18.452.
Coffin-Lowry Syndrome	C16.320.400.525.249	C18.452. C10.597.	C19.53. C16.320.
Fragile X Syndrome	C16.320.400.525.500	C10.597. C16.320.	C16.131. C16.320.
Lesch-Nyhan Syndrome	C16.320.400.525.625	C10.228. C10.597. C16.320. C16.320. C16.320. C18.452. C18.452.	C10.574. C16.320. C16.320. C18.452. C18.452.
Menkes Kinky Hair Syndrome	C16.320.400.525.687	C10.228. C10.597. C16.320. C16.320. C16.320. C18.452. C18.452.	C10.574. C16.320. C16.320. C17.800. C18.452.
Mucopolysaccharidosis II	C16.320.400.525.750	C10.597. C16.320. C17.300. C18.452.	C16.320. C16.320. C18.452.
Pyruvate Dehydrogenase Complex Deficiency Disease	C16.320.400.525.875	C10.228. C16.320. C16.320. C18.452. C18.452.	C10.597. C16.320. C18.452.
Rett Syndrome	C16.320.400.525.937	C10.574. C16.320. F3.550.	C10.597. C16.320.
Myotonia Congenita	C16.320.400.540	C5.651. C10.668.	C10.574.
Myotonic Dystrophy	C16.320.400.542	C5.651. C10.574. C10.668.	C5.651. C10.668. C16.320.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Hereditary Degenerative Disorders, Nervous System
Neuroacanthocytosis

Neuroacanthocytosis	C16.320.400.550	C10.228.	
Neurofibromatoses	C16.320.400.560	C4.557.	C4.700.
		C10.562.	C10.574.
		C16.320.	
Neurofibromatosis 1	C16.320.400.560.400	C4.557.	C4.700.
		C10.562.	C10.574.
		C10.668.	C16.320.
Neurofibromatosis 2	C16.320.400.560.700	C4.557.	C4.557.
		C4.557.	C4.588.
		C4.700.	C9.218.
		C9.647.	C10.292.
		C10.292.	C10.551.
		C10.562.	C10.574.
		C16.320.	
Neuronal Ceroid-Lipofuscinoses	C16.320.400.600	C10.574.	C16.320.
		C18.452.	C18.452.
Optic Atrophies, Hereditary	C16.320.400.630	C10.292.	C10.574.
		C11.270.	C11.640.
		C16.320.	
Optic Atrophy, Autosomal Dominant	C16.320.400.630.100	C10.292.	C10.574.
		C11.270.	C11.640.
		C16.320.	C18.452.
Optic Atrophy, Hereditary, Leber	C16.320.400.630.400	C10.292.	C10.574.
		C11.270.	C11.640.
		C16.320.	C18.452.
Wolfram Syndrome	C16.320.400.630.980	C9.218.	C10.292.
		C10.574.	C10.597.
		C10.597.	C11.270.
		C11.640.	C11.966.
		C12.777.	C13.351.
		C16.131.	C16.131.
		C16.320.	C18.452.
		C19.246.	C19.700.
Rett Syndrome	C16.320.400.700	C10.574.	C10.597.
		C16.320.	C16.320.
		F3.550.	
Spinal Muscular Atrophies of Childhood	C16.320.400.765	C10.228.	C10.574.
		C10.574.	C10.668.
		C10.668.	
Spinocerebellar Degenerations	C16.320.400.780	C10.228.	C10.228.
		C10.574.	
Friedreich Ataxia	C16.320.400.780.200	C10.228.	C10.228.
		C10.574.	C18.452.
Myoclonic Cerebellar Dyssynergia	C16.320.400.780.500	C10.228.	C10.228.
		C10.574.	
Olivopontocerebellar Atrophies	C16.320.400.780.750	C10.228.	C10.228.
		C10.228.	C10.228.
		C10.574.	C10.574.
		C10.574.	
Spinocerebellar Ataxias	C16.320.400.780.875	C10.228.	C10.228.
		C10.228.	C10.574.
		C10.597.	
Machado-Joseph Disease	C16.320.400.780.875.500	C10.228.	C10.228.
		C10.228.	C10.574.
		C10.597.	
Tourette Syndrome	C16.320.400.820	C10.228.	C10.228.
		C10.574.	F3.550.
Tuberous Sclerosis	C16.320.400.880	C4.445.	C10.500.
		C10.562.	C10.574.
		C16.131.	
Unverricht-Lundborg Syndrome	C16.320.400.940	C10.228.	C10.574.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Hyperthyroxinemia, Familial Dysalbuminemic

Hyperthyroxinemia, Familial Dysalbuminemic	C16.320.427	C19.874.	
Kallmann Syndrome	C16.320.467	C12.706.	C13.351.
		C16.131.	C19.391.
		C19.391.	
Kartagener Syndrome	C16.320.480	C8.127.	C8.200.
		C8.695.	C9.150.
		C14.240.	C14.280.
		C16.131.	C16.131.
		C16.131.	
Marfan Syndrome	C16.320.540	C5.116.	C14.240.
		C14.280.	C16.131.
		C16.131.	C17.300.
Metabolism, Inborn Errors	C16.320.565	C18.452.	
Amino Acid Metabolism, Inborn Errors	C16.320.565.100	C18.452.	
Albinism	C16.320.565.100.102	C11.270.	C16.320.
		C16.320.	C17.800.
		C17.800.	C18.452.
Albinism, Ocular	C16.320.565.100.102.90	C11.270.	C16.320.
		C16.320.	C17.800.
		C17.800.	C18.452.
Albinism, Oculocutaneous	C16.320.565.100.102.100	C11.270.	C16.320.
		C16.320.	C17.800.
		C17.800.	C18.452.
Hermanski-Pudlak Syndrome	C16.320.565.100.102.100.400	C11.270.	C15.378.
		C15.378.	C15.378.
		C15.378.	C16.320.
		C16.320.	C16.320.
		C17.800.	C17.800.
		C18.452.	
Piebaldism	C16.320.565.100.102.600	C16.320.	C16.320.
		C17.800.	C17.800.
		C18.452.	
Alkaptonuria	C16.320.565.100.187	C18.452.	
Carbamoyl-Phosphate Synthase I Deficiency Disease	C16.320.565.100.275	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
Citrullinemia	C16.320.565.100.340	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Homocystinuria	C16.320.565.100.470	C10.228.	C16.320.
		C17.300.	C18.452.
		C18.452.	C18.452.
Hyperargininemia	C16.320.565.100.475	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Hyperglycinemia, Nonketotic	C16.320.565.100.477	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Hyperhomocysteinemia	C16.320.565.100.480	C18.452.	
Hyperlysinemias	C16.320.565.100.544	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Maple Syrup Urine Disease	C16.320.565.100.608	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Multiple Acyl Coenzyme A Dehydrogenase Deficiency	C16.320.565.100.614	C18.452.	C18.452.
Multiple Carboxylase Deficiency	C16.320.565.100.620	C16.320.	C18.452.
		C18.452.	
Biotinidase Deficiency	C16.320.565.100.620.100	C16.320.	C18.452.
		C18.452.	

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Amino Acid Metabolism, Inborn Errors

Multiple Carboxylase Deficiency

Holocarboxylase Synthetase Deficiency

Holocarboxylase Synthetase Deficiency

C16.320.565.100.620.380

C16.320.
C18.452.

C18.452.

Ornithine Carbamoyltransferase Deficiency Disease

C16.320.565.100.729

C10.228.
C18.452.
C18.452.

C16.320.
C18.452.

Phenylketonurias

C16.320.565.100.766

C10.228.
C18.452.
C18.452.

C16.320.
C18.452.

Phenylketonuria, Maternal

C16.320.565.100.766.500

C10.228.
C16.320.
C18.452.

C13.703.
C18.452.

Tyrosinemias

C16.320.565.100.880

C10.228.
C18.452.
C18.452.

C16.320.
C18.452.

Amino Acid Transport Disorders, Inborn

Hartnup Disease

C16.320.565.151

C16.320.565.151.355

C18.452.
C10.228.
C13.351.

C12.777.
C16.320.

Oculocerebrorenal Syndrome

C16.320.565.151.600

C16.320.
C18.452.
C10.228.

C12.777.
C16.131.

Amyloidosis, Familial

Amyloid Neuropathies, Familial

C16.320.565.176

C16.320.565.176.50

C18.452.
C10.574.
C16.320.

C18.452.
C10.668.
C18.452.

Cerebral Amyloid Angiopathy, Familial

C16.320.565.176.160

C18.452.
C10.228.
C14.907.

C10.228.
C16.320.
C18.452.

Brain Diseases, Metabolic, Inborn

Carbamoyl-Phosphate Synthase I Deficiency Disease

C16.320.565.189

C16.320.565.189.162

C10.228.
C18.452.
C18.452.

C18.452.
C16.320.
C18.452.

Cerebral Amyloid Angiopathy, Familial

C16.320.565.189.168

C10.228.
C14.907.
C18.452.

C10.228.
C16.320.
C18.452.

Citrullinemia

C16.320.565.189.175

C18.452.
C10.228.
C18.452.

C16.320.
C18.452.

Galactosemias

C16.320.565.189.320

C18.452.
C10.228.
C18.452.

C18.452.
C16.320.
C18.452.

Hartnup Disease

C16.320.565.189.355

C18.452.
C10.228.
C13.351.

C12.777.
C16.320.
C18.452.

Hepatolenticular Degeneration

C16.320.565.189.360

C18.452.
C10.228.
C10.574.

C10.228.
C16.320.
C18.452.

C16.320.
C18.452.
C6.552.

C10.228.
C10.228.
C16.320.

C10.574.
C16.320.
C18.452.

C10.228.
C16.320.
C18.452.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Brain Diseases, Metabolic, Inborn

Homocystinuria

Homocystinuria	C16.320.565.189.365	C10.228. C17.300. C18.452.	C16.320. C18.452.
Hyperargininemia	C16.320.565.189.370	C10.228. C18.452.	C16.320. C18.452.
Hyperglycinemia, Nonketotic	C16.320.565.189.375	C10.228. C18.452.	C16.320. C18.452.
Hyperlysinemias	C16.320.565.189.380	C10.228. C18.452.	C16.320. C18.452.
Leigh Disease	C16.320.565.189.412	C10.228. C18.452.	C16.320. C18.452.
Lesch-Nyhan Syndrome	C16.320.565.189.425	C10.228. C10.597. C16.320. C16.320.	C10.574. C16.320. C18.452.
Lysosomal Storage Diseases, Nervous System	C16.320.565.189.435	C10.228. C18.452.	C16.320. C18.452.
Fucosidosis	C16.320.565.189.435.295	C10.228. C16.320. C18.452.	C16.320. C18.452.
Glycogen Storage Disease Type II	C16.320.565.189.435.340	C10.228. C16.320. C18.452.	C16.320. C18.452.
Mucopolidoses	C16.320.565.189.435.590	C5.116. C16.320. C18.452.	C10.228. C16.320. C18.452.
Sialic Acid Storage Disease	C16.320.565.189.435.810	C10.228. C18.452.	C16.320. C18.452.
Sphingolipidoses	C16.320.565.189.435.825	C10.228. C16.320. C18.452.	C16.320. C18.452.
Fabry Disease	C16.320.565.189.435.825.200	C10.228. C16.320. C18.452.	C16.320. C18.452.
Gangliosidoses	C16.320.565.189.435.825.300	C10.228. C16.320. C18.452.	C16.320. C18.452.
Gangliosidoses, GM2	C16.320.565.189.435.825.300.300	C10.228. C16.320. C18.452.	C16.320. C18.452.
Sandhoff Disease	C16.320.565.189.435.825.300.300.249	C10.228. C16.320. C16.320.	C10.228. C16.320. C18.452.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Brain Diseases, Metabolic, Inborn

Lysosomal Storage Diseases, Nervous System

Sphingolipidoses

Tay-Sachs Disease	C16.320.565.189.435.825.300.300.500	C10.228. C16.320. C18.452. C18.452. C18.452.	C16.320. C18.452. C18.452.
Tay-Sachs Disease, AB Variant	C16.320.565.189.435.825.300.300.750	C10.228. C16.320. C18.452. C18.452. C18.452.	C16.320. C18.452. C18.452.
Gangliosidosis, GM1	C16.320.565.189.435.825.300.400	C10.228. C16.320. C18.452. C18.452. C18.452.	C16.320. C18.452. C18.452.
Sandhoff Disease	C16.320.565.189.435.825.300.700	C10.228. C16.320. C16.320. C18.452. C18.452. C18.452.	C10.228. C16.320. C18.452. C18.452.
Gaucher Disease	C16.320.565.189.435.825.400	C10.228. C16.320. C18.452. C18.452. C18.452.	C15.604. C16.320. C18.452. C18.452.
Leukodystrophy, Globoid Cell	C16.320.565.189.435.825.590	C10.228. C16.320. C18.452. C18.452. C18.452.	C10.314. C16.320. C18.452. C18.452.
Niemann-Pick Diseases	C16.320.565.189.435.825.700	C10.228. C16.320. C18.452. C18.452. C18.452.	C15.604. C16.320. C18.452. C18.452.
Niemann-Pick Disease, Type A	C16.320.565.189.435.825.700.500	C10.228. C16.320. C18.452. C18.452. C18.452.	C15.604. C16.320. C18.452. C18.452.
Niemann-Pick Disease, Type B	C16.320.565.189.435.825.700.750	C10.228. C16.320. C18.452. C18.452. C18.452.	C15.604. C16.320. C18.452. C18.452.
Niemann-Pick Disease, Type C	C16.320.565.189.435.825.700.875	C10.228. C16.320. C18.452. C18.452. C18.452.	C15.604. C16.320. C18.452. C18.452.
Sea-Blue Histiocyte Syndrome	C16.320.565.189.435.825.775	C10.228. C16.320. C18.452. C18.452. C18.452.	C15.604. C16.320. C18.452. C18.452.
Sulfatidosis	C16.320.565.189.435.825.850	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452. C18.452.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Brain Diseases, Metabolic, Inborn

Lysosomal Storage Diseases, Nervous System

Sphingolipidoses

Leukodystrophy, Metachromatic	C16.320.565.189.435.825.850.500	C10.228. C16.320. C18.452. C18.452. C18.452.	C10.314. C16.320. C18.452. C18.452.
Multiple Sulfatase Deficiency Disease	C16.320.565.189.435.825.850.750	C10.228. C16.320. C18.452. C18.452. C18.452.	C16.320. C18.452. C18.452. C18.452.
Maple Syrup Urine Disease	C16.320.565.189.520	C10.228. C18.452. C18.452.	C16.320. C18.452.
MELAS Syndrome	C16.320.565.189.535	C5.651. C10.668. C18.452.	C10.228. C18.452. C18.452.
Menkes Kinky Hair Syndrome	C16.320.565.189.540	C10.228. C10.597. C16.320. C16.320. C18.452. C18.452.	C10.574. C16.320. C16.320. C17.800. C18.452.
MERRF Syndrome	C16.320.565.189.545	C5.651. C10.228. C18.452. C18.452.	C10.228. C10.668. C18.452.
Oculocerebrorenal Syndrome	C16.320.565.189.640	C10.228. C13.351. C16.320. C16.320. C18.452. C18.452.	C12.777. C16.131. C16.320. C18.452. C18.452.
Ornithine Carbamoyltransferase Deficiency Disease	C16.320.565.189.650	C10.228. C18.452. C18.452.	C16.320. C18.452.
Peroxisomal Disorders	C16.320.565.189.680	C10.228. C18.452. C18.452.	C16.320. C18.452.
Adrenoleukodystrophy	C16.320.565.189.680.100	C10.228. C10.597. C16.320. C18.452. C18.452.	C10.314. C16.320. C16.320. C18.452.
Mevalonate Kinase Deficiency	C16.320.565.189.680.430	C10.228. C16.320. C18.452. C20.683.	C15.378. C18.452. C18.452.
Refsum Disease	C16.320.565.189.680.760	C10.228. C10.574. C16.131. C16.320. C18.452.	C10.500. C10.668. C16.320. C18.452.
Refsum Disease, Infantile	C16.320.565.189.680.865	C10.228. C18.452. C18.452.	C16.320. C18.452.
Zellweger Syndrome	C16.320.565.189.680.970	C6.552. C12.777. C16.131. C18.452. C18.452.	C10.228. C13.351. C16.320. C18.452.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Brain Diseases, Metabolic, Inborn

Phenylketonurias

Phenylketonurias	C16.320.565.189.687	C10.228. C18.452. C18.452.	C16.320. C18.452.
Phenylketonuria, Maternal	C16.320.565.189.687.500	C10.228. C16.320. C18.452.	C13.703. C18.452. C18.452.
Pyruvate Carboxylase Deficiency Disease	C16.320.565.189.725	C10.228. C18.452. C18.452.	C16.320. C18.452. C18.452.
Pyruvate Dehydrogenase Complex Deficiency Disease	C16.320.565.189.750	C10.228. C16.320. C16.320. C18.452. C18.452.	C10.597. C16.320. C18.452. C18.452.
Tyrosinemias	C16.320.565.189.875	C10.228. C18.452. C18.452.	C16.320. C18.452.
Carbohydrate Metabolism, Inborn Errors	C16.320.565.202	C18.452.	
Carbohydrate-Deficient Glycoprotein Syndrome	C16.320.565.202.125	C18.452.	
Fructose Metabolism, Inborn Errors	C16.320.565.202.251	C18.452.	
Fructose-1,6-Diphosphatase Deficiency	C16.320.565.202.251.221	C18.452.	
Fructose Intolerance	C16.320.565.202.251.271	C18.452.	
Fucosidosis	C16.320.565.202.303	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452. C18.452.
Galactosemias	C16.320.565.202.355	C10.228. C18.452. C18.452.	C16.320. C18.452.
Glucosephosphate Dehydrogenase Deficiency	C16.320.565.202.402	C15.378. C18.452.	C16.320.
Glycogen Storage Disease	C16.320.565.202.449	C18.452.	
Glycogen Storage Disease Type I	C16.320.565.202.449.448	C18.452.	
Glycogen Storage Disease Type II	C16.320.565.202.449.500	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452.
Glycogen Storage Disease Type IIb	C16.320.565.202.449.510	C10.597. C16.320.	C14.280. C18.452.
Glycogen Storage Disease Type III	C16.320.565.202.449.520	C18.452.	
Glycogen Storage Disease Type IV	C16.320.565.202.449.540	C18.452.	
Glycogen Storage Disease Type V	C16.320.565.202.449.560	C18.452.	
Glycogen Storage Disease Type VI	C16.320.565.202.449.580	C18.452.	
Glycogen Storage Disease Type VII	C16.320.565.202.449.600	C5.651. C18.452.	C16.320.
Glycogen Storage Disease Type VIII	C16.320.565.202.449.620	C16.320.	C18.452.
Hyperoxaluria, Primary	C16.320.565.202.460	C12.777. C18.452.	C13.351.
Lactose Intolerance	C16.320.565.202.589	C6.405. C18.452.	C18.452.
Mannosidase Deficiency Diseases	C16.320.565.202.607	C16.320. C18.452.	C18.452.
alpha-Mannosidosis	C16.320.565.202.607.500	C16.320. C18.452.	C18.452.
beta-Mannosidosis	C16.320.565.202.607.750	C16.320. C18.452.	C18.452.
Mucopolipidoses	C16.320.565.202.670	C5.116. C16.320. C18.452. C18.452.	C10.228. C16.320. C18.452. C18.452.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Carbohydrate Metabolism, Inborn Errors

Mucopolysaccharidoses

Mucopolysaccharidoses	C16.320.565.202.715	C16.320. C18.452.	C17.300. C18.452.
Mucopolysaccharidosis I	C16.320.565.202.715.640	C16.320. C18.452.	C17.300. C18.452.
Mucopolysaccharidosis II	C16.320.565.202.715.645	C10.597. C16.320. C17.300. C18.452.	C16.320. C16.320. C18.452.
Mucopolysaccharidosis III	C16.320.565.202.715.650	C16.320. C18.452.	C17.300. C18.452.
Mucopolysaccharidosis IV	C16.320.565.202.715.655	C16.320. C18.452.	C17.300. C18.452.
Mucopolysaccharidosis VI	C16.320.565.202.715.670	C16.320. C18.452.	C17.300. C18.452.
Mucopolysaccharidosis VII	C16.320.565.202.715.675	C16.320. C18.452.	C17.300. C18.452.
Multiple Carboxylase Deficiency	C16.320.565.202.720	C16.320. C18.452.	C18.452.
Biotinidase Deficiency	C16.320.565.202.720.100	C16.320. C18.452.	C18.452.
Holocarboxylase Synthetase Deficiency	C16.320.565.202.720.380	C16.320. C18.452.	C18.452.
Pyruvate Metabolism, Inborn Errors	C16.320.565.202.810	C18.452.	
Leigh Disease	C16.320.565.202.810.444	C10.228. C18.452.	C16.320. C18.452.
Pyruvate Carboxylase Deficiency Disease	C16.320.565.202.810.666	C10.228. C18.452. C18.452.	C16.320. C18.452.
Pyruvate Dehydrogenase Complex Deficiency Disease	C16.320.565.202.810.766	C10.228. C16.320. C16.320. C18.452. C18.452.	C10.597. C16.320. C16.320. C18.452. C18.452.
Cytochrome-c Oxidase Deficiency	C16.320.565.240	C18.452.	
Hyperbilirubinemia, Hereditary	C16.320.565.300	C18.452.	
Crigler-Najjar Syndrome	C16.320.565.300.281	C18.452.	
Gilbert Disease	C16.320.565.300.528	C18.452.	
Jaundice, Chronic Idiopathic	C16.320.565.300.764	C16.614.	C18.452.
Lipid Metabolism, Inborn Errors	C16.320.565.398	C18.452.	C18.452.
Hyperlipidemia, Familial Combined	C16.320.565.398.450	C18.452.	C18.452.
Hyperlipoproteinemia Type I	C16.320.565.398.465	C18.452.	C18.452.
Hyperlipoproteinemia Type II	C16.320.565.398.481	C18.452.	C18.452.
Hyperlipoproteinemia Type III	C16.320.565.398.483	C18.452.	C18.452.
Hyperlipoproteinemia Type IV	C16.320.565.398.487	C18.452. C18.452.	C18.452.
Hyperlipoproteinemia Type V	C16.320.565.398.493	C18.452.	C18.452.
Hypolipoproteinemias	C16.320.565.398.500	C18.452.	C18.452.
Hypoalphalipoproteinemias	C16.320.565.398.500.330	C18.452.	C18.452.
Lecithin Acyltransferase Deficiency	C16.320.565.398.500.330.500	C18.452.	C18.452.
Tangier Disease	C16.320.565.398.500.330.750	C10.668. C18.452.	C18.452.
Hypobetalipoproteinemias	C16.320.565.398.500.440	C18.452.	C18.452.
Abetalipoproteinemia	C16.320.565.398.500.440.500	C18.452.	C18.452.
Lipidoses	C16.320.565.398.641	C18.452.	C18.452.
Cholesterol Ester Storage Disease	C16.320.565.398.641.201	C16.320. C18.452.	C18.452. C18.452.
Wolman Disease	C16.320.565.398.641.201.500	C16.320. C18.452.	C16.614. C18.452.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Lipid Metabolism, Inborn Errors

Lipidoses

Neuronal Ceroid-Lipofuscinoses

Neuronal Ceroid-Lipofuscinoses

Sjogren-Larsson Syndrome

Sphingolipidoses

Fabry Disease

Gangliosidoses

Gangliosidoses, GM2

Sandhoff Disease

Tay-Sachs Disease

Tay-Sachs Disease, AB Variant

Gangliosidosis, GM1

Gaucher Disease

Leukodystrophy, Globoid Cell

Niemann-Pick Diseases

Niemann-Pick Disease, Type A

C16.320.565.398.641.509	C10.574. C18.452.	C16.320. C18.452.
C16.320.565.398.641.723	C16.131. C16.614. C17.800. C17.800.	C16.320. C17.800. C17.800.
C16.320.565.398.641.803	C18.452. C10.228. C16.320. C18.452.	C18.452. C16.320. C18.452.
C16.320.565.398.641.803.300	C18.452. C10.228. C16.320. C18.452. C18.452.	C18.452. C16.320. C16.320. C18.452.
C16.320.565.398.641.803.350	C18.452. C10.228. C16.320. C18.452.	C18.452. C16.320. C18.452.
C16.320.565.398.641.803.350.300	C18.452. C10.228. C16.320. C18.452. C18.452.	C18.452. C16.320. C18.452.
C16.320.565.398.641.803.350.300.700	C18.452. C10.228. C16.320. C18.452. C18.452.	C18.452. C10.228. C16.320. C18.452.
C16.320.565.398.641.803.350.300.850	C18.452. C10.228. C16.320. C18.452.	C18.452. C16.320. C18.452.
C16.320.565.398.641.803.350.300.925	C18.452. C10.228. C16.320. C18.452.	C18.452. C16.320. C18.452.
C16.320.565.398.641.803.350.360	C18.452. C10.228. C16.320. C18.452.	C18.452. C16.320. C18.452.
C16.320.565.398.641.803.441	C18.452. C10.228. C16.320. C18.452.	C18.452. C15.604. C16.320. C18.452.
C16.320.565.398.641.803.585	C18.452. C10.228. C16.320. C18.452. C18.452.	C18.452. C10.314. C16.320. C18.452.
C16.320.565.398.641.803.730	C18.452. C10.228. C16.320. C18.452. C18.452.	C18.452. C15.604. C16.320. C18.452.
C16.320.565.398.641.803.730.500	C18.452. C10.228. C16.320. C18.452. C18.452.	C18.452. C15.604. C16.320. C18.452.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Lipid Metabolism, Inborn Errors

Lipidoses

Sphingolipidoses

Niemann-Pick Disease, Type B	C16.320.565.398.641.803.730.750	C10.228. C16.320. C18.452. C18.452. C18.452.	C15.604. C16.320. C18.452. C18.452.
Niemann-Pick Disease, Type C	C16.320.565.398.641.803.730.875	C10.228. C16.320. C18.452. C18.452. C18.452.	C15.604. C16.320. C18.452. C18.452.
Sea-Blue Histiocyte Syndrome	C16.320.565.398.641.803.850	C10.228. C16.320. C18.452. C18.452. C18.452.	C15.604. C16.320. C18.452. C18.452.
Sulfatidosis	C16.320.565.398.641.803.925	C10.228. C16.320. C18.452. C18.452. C18.452.	C16.320. C18.452. C18.452.
Leukodystrophy, Metachromatic	C16.320.565.398.641.803.925.500	C10.228. C16.320. C18.452. C18.452. C18.452.	C10.314. C16.320. C18.452. C18.452.
Multiple Sulfatase Deficiency Disease	C16.320.565.398.641.803.925.750	C10.228. C16.320. C18.452. C18.452. C18.452.	C16.320. C18.452. C18.452.
Smith-Lemli-Opitz Syndrome	C16.320.565.398.850	C16.131. C18.452. C18.452. C18.452.	C16.320. C18.452.
Xanthomatosis, Cerebrotendinous	C16.320.565.398.925	C18.452.	C18.452.
Lysosomal Storage Diseases	C16.320.565.595	C18.452.	
Cholesterol Ester Storage Disease	C16.320.565.595.201	C16.320. C18.452. C18.452.	C18.452.
Wolman Disease	C16.320.565.595.201.500	C16.320. C18.452. C18.452.	C16.614. C18.452.
Cystinosis	C16.320.565.595.377	C18.452.	
Lysosomal Storage Diseases, Nervous System	C16.320.565.595.554	C10.228. C18.452. C18.452.	C16.320. C18.452.
Fucosidosis	C16.320.565.595.554.295	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452.
Glycogen Storage Disease Type II	C16.320.565.595.554.340	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452.
Mucopolidoses	C16.320.565.595.554.590	C5.116. C16.320. C18.452. C18.452.	C10.228. C16.320. C18.452.
Sialic Acid Storage Disease	C16.320.565.595.554.810	C10.228. C18.452. C18.452.	C16.320. C18.452.
Sphingolipidoses	C16.320.565.595.554.825	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452. C18.452.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Lysosomal Storage Diseases

Lysosomal Storage Diseases, Nervous System

Sphingolipidoses

Fabry Disease	C16.320.565.595.554.825.200	C10.228. C16.320. C18.452. C18.452. C18.452.	C16.320. C16.320. C18.452. C18.452.
Gangliosidoses	C16.320.565.595.554.825.300	C10.228. C16.320. C18.452. C18.452. C18.452.	C16.320. C18.452. C18.452.
Gangliosidoses, GM2	C16.320.565.595.554.825.300.300	C10.228. C16.320. C18.452. C18.452. C18.452.	C16.320. C18.452.
Sandhoff Disease	C16.320.565.595.554.825.300.300.800	C10.228. C16.320. C16.320. C18.452. C18.452. C18.452. C18.452.	C10.228. C16.320. C18.452. C18.452.
Tay-Sachs Disease	C16.320.565.595.554.825.300.300.840	C10.228. C16.320. C18.452. C18.452. C18.452.	C16.320. C18.452. C18.452.
Tay-Sachs Disease, AB Variant	C16.320.565.595.554.825.300.300.920	C10.228. C16.320. C18.452. C18.452. C18.452.	C16.320. C18.452. C18.452.
Gangliosidosis, GM1	C16.320.565.595.554.825.300.400	C10.228. C16.320. C18.452. C18.452. C18.452.	C16.320. C18.452. C18.452.
Gaucher Disease	C16.320.565.595.554.825.400	C10.228. C16.320. C18.452. C18.452. C18.452.	C15.604. C16.320. C18.452. C18.452.
Leukodystrophy, Globoid Cell	C16.320.565.595.554.825.590	C10.228. C16.320. C18.452. C18.452. C18.452.	C10.314. C16.320. C18.452. C18.452.
Niemann-Pick Diseases	C16.320.565.595.554.825.700	C10.228. C16.320. C18.452. C18.452. C18.452.	C15.604. C16.320. C18.452. C18.452.
Niemann-Pick Disease, Type A	C16.320.565.595.554.825.700.500	C10.228. C16.320. C18.452. C18.452. C18.452.	C15.604. C16.320. C18.452. C18.452.
Niemann-Pick Disease, Type B	C16.320.565.595.554.825.700.750	C10.228. C16.320. C18.452. C18.452. C18.452.	C15.604. C16.320. C18.452. C18.452.
Niemann-Pick Disease, Type C	C16.320.565.595.554.825.700.875	C10.228. C16.320. C18.452. C18.452. C18.452.	C15.604. C16.320. C18.452. C18.452.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Lysosomal Storage Diseases

Lysosomal Storage Diseases, Nervous System

Sphingolipidoses

Sea-Blue Histiocyte Syndrome	C16.320.565.595.554.825.775	C10.228. C16.320. C18.452. C18.452. C18.452.	C15.604. C16.320. C18.452. C18.452.
Sulfatidosis	C16.320.565.595.554.825.850	C10.228. C16.320. C18.452. C18.452. C18.452.	C16.320. C18.452. C18.452.
Leukodystrophy, Metachromatic	C16.320.565.595.554.825.850.500	C10.228. C16.320. C18.452. C18.452. C18.452.	C10.314. C16.320. C18.452. C18.452.
Multiple Sulfatase Deficiency Disease	C16.320.565.595.554.825.850.750	C10.228. C16.320. C18.452. C18.452. C18.452.	C16.320. C18.452. C18.452.
Mannosidase Deficiency Diseases	C16.320.565.595.577	C16.320. C18.452.	C18.452.
alpha-Mannosidosis	C16.320.565.595.577.500	C16.320. C18.452.	C18.452.
beta-Mannosidosis	C16.320.565.595.577.750	C16.320. C18.452.	C18.452.
Mucopolysaccharidoses	C16.320.565.595.600	C16.320. C18.452.	C17.300. C18.452.
Mucopolysaccharidosis I	C16.320.565.595.600.640	C16.320. C18.452.	C17.300. C18.452.
Mucopolysaccharidosis II	C16.320.565.595.600.645	C10.597. C16.320. C17.300. C18.452.	C16.320. C16.320. C18.452.
Mucopolysaccharidosis III	C16.320.565.595.600.650	C16.320. C18.452.	C17.300. C18.452.
Mucopolysaccharidosis IV	C16.320.565.595.600.655	C16.320. C18.452.	C17.300. C18.452.
Mucopolysaccharidosis VI	C16.320.565.595.600.670	C16.320. C18.452.	C17.300. C18.452.
Mucopolysaccharidosis VII	C16.320.565.595.600.675	C16.320. C18.452.	C17.300. C18.452.
Metal Metabolism, Inborn Errors	C16.320.565.618	C18.452.	
Hemochromatosis	C16.320.565.618.337	C18.452.	C18.452.
Hepatolenticular Degeneration	C16.320.565.618.403	C6.552. C10.228. C10.574. C16.320. C18.452. C18.452.	C10.228. C10.228. C16.320. C18.452. C18.452.
Hypophosphatasia	C16.320.565.618.482	C18.452.	
Hypophosphatemia, Familial	C16.320.565.618.544	C12.777. C16.320. C18.452. C18.452.	C13.351. C18.452. C18.452.
Hypophosphatemic Rickets, X-Linked Dominant	C16.320.565.618.544.500	C5.116. C13.351. C16.320. C18.452. C18.654.	C12.777. C16.320. C18.452.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Metal Metabolism, Inborn Errors

Menkes Kinky Hair Syndrome

Menkes Kinky Hair Syndrome	C16.320.565.618.590	C10.228. C10.597. C16.320. C16.320. C18.452. C18.452.	C10.574. C16.320. C16.320. C17.800. C18.452.
Paralyses, Familial Periodic	C16.320.565.618.711	C5.651. C18.452.	C10.668.
Hypokalemic Periodic Paralysis	C16.320.565.618.711.550	C5.651. C18.452.	C10.668.
Paralysis, Hyperkalemic Periodic	C16.320.565.618.711.600	C5.651. C18.452.	C10.668.
Pseudohypoparathyroidism	C16.320.565.618.815	C5.116. C18.452.	C18.452.
Pseudopseudohypoparathyroidism	C16.320.565.618.815.815	C5.116. C18.452.	C18.452.
Peroxisomal Disorders	C16.320.565.663	C10.228. C18.452. C18.452.	C16.320. C18.452.
Acatalsia	C16.320.565.663.25	C18.452.	
Adrenoleukodystrophy	C16.320.565.663.112	C10.228. C10.597. C16.320. C18.452. C18.452.	C10.314. C16.320. C16.320. C18.452.
Chondrodysplasia Punctata, Rhizomelic	C16.320.565.663.200	C5.116.	C18.452.
Mevalonate Kinase Deficiency	C16.320.565.663.480	C10.228. C16.320. C18.452.	C15.378. C18.452.
Refsum Disease	C16.320.565.663.760	C20.683. C10.228. C10.574. C16.131. C16.320.	C10.500. C10.668. C16.320. C18.452.
Refsum Disease, Infantile	C16.320.565.663.865	C18.452. C10.228. C18.452. C18.452.	C18.452. C16.320. C18.452.
Zellweger Syndrome	C16.320.565.663.970	C6.552. C12.777. C16.131. C18.452. C18.452.	C10.228. C13.351. C16.320. C18.452.
Porphyrias	C16.320.565.708	C18.452.	C18.452.
Porphyria, Erythropoietic	C16.320.565.708.250	C17.800. C18.452. C16.320. C17.800. C18.452.	C18.452. C18.452. C17.800. C18.452.
Porphyrias, Hepatic	C16.320.565.708.400	C6.552. C17.800. C18.452. C18.452.	C16.320. C17.800. C18.452.
Coproporphyrin, Hereditary	C16.320.565.708.400.74	C18.452. C6.552. C17.800. C18.452.	C16.320. C17.800. C18.452.
Porphyria, Acute Intermittent	C16.320.565.708.400.150	C6.552. C17.800. C18.452. C18.452.	C16.320. C17.800. C18.452.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Porphyrias

Porphyrias, Hepatic

Porphyria Cutanea Tarda

Porphyria Cutanea Tarda

	C16.320.565.708.400.250		C6.552. C17.800. C18.452. C18.452.	C16.320. C17.800. C18.452.
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Porphyria, Hepatoerythropoietic

	C16.320.565.708.400.437		C6.552. C17.800. C18.452. C18.452.	C16.320. C17.800. C18.452.
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Porphyria, Variegata

	C16.320.565.708.400.625		C6.552. C17.800. C18.452. C18.452.	C16.320. C17.800. C18.452.
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Protoporphyrin, Erythropoietic

	C16.320.565.708.400.812		C6.552. C17.800. C18.452. C18.452.	C16.320. C17.800. C18.452.
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Progeria

	C16.320.565.753		C18.452.	
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Purine-Pyrimidine Metabolism, Inborn Errors

	C16.320.565.798		C18.452.	
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Dihydropyrimidine Dehydrogenase Deficiency

	C16.320.565.798.183		C18.452.	
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Gout

	C16.320.565.798.368		C5.550. C18.452.	C5.799.
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Arthritis, Gouty

	C16.320.565.798.368.410		C5.550. C18.452.	C5.799.
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Lesch-Nyhan Syndrome

	C16.320.565.798.594		C10.228. C10.597. C16.320. C16.320. C18.452.	C10.574. C16.320. C16.320. C18.452.
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Renal Tubular Transport, Inborn Errors

	C16.320.565.861		C12.777. C18.452.	C13.351.
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Acidosis, Renal Tubular

	C16.320.565.861.93		C12.777. C18.452.	C13.351. C18.452.
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Fanconi Syndrome

	C16.320.565.861.450		C12.777. C13.351. C18.452.	C12.777. C13.351.
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Glycosuria, Renal

	C16.320.565.861.532		C12.777. C13.351. C18.452.	C12.777. C13.351. C18.452.
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Hypophosphatemia, Familial

	C16.320.565.861.647		C12.777. C16.320. C18.452.	C13.351. C18.452.
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Hypophosphatemic Rickets, X-Linked Dominant

	C16.320.565.861.647.500		C5.116. C13.351. C16.320. C18.452.	C12.777. C16.320. C18.452.
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Oculocerebrorenal Syndrome

	C16.320.565.861.750		C10.228. C13.351. C16.320. C16.320. C18.452. C18.452.	C12.777. C16.131. C16.320. C18.452.
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Pseudohypoaldosteronism

	C16.320.565.861.770		C12.777. C18.452.	C13.351. C19.53.
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Renal Aminoacidurias

	C16.320.565.861.885		C12.777. C18.452.	C13.351.
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Cystinuria

	C16.320.565.861.885.250		C12.777. C18.452.	C13.351.
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C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Renal Tubular Transport, Inborn Errors

Renal Aminoacidurias

Hartnup Disease

Hartnup Disease

C16.320.565.861.885.457

C10.228.
C12.777.
C13.351.
C16.320.
C18.452.
C18.452.

Steroid Metabolism, Inborn Errors

Adrenal Hyperplasia, Congenital

C16.320.565.925
C16.320.565.925.249

C18.452.
C16.320.
C18.452.
C19.53.

Ichthyosis, X-Linked

C16.320.565.925.400

C16.131.
C16.320.
C17.800.
C17.800.
C18.452.

**Mineralocorticoid Excess Syndrome, Apparent
Smith-Lemli-Opitz Syndrome**

C16.320.565.925.500
C16.320.565.925.875

C18.452.
C16.131.
C18.452.
C18.452.

Muscular Dystrophies

Distal Myopathies

Glycogen Storage Disease Type VII

C16.320.577
C16.320.577.74
C16.320.577.149

C5.651.
C5.651.
C5.651.
C18.452.

Muscular Dystrophies, Limb-Girdle

Muscular Dystrophy, Duchenne

C16.320.577.280
C16.320.577.300

C5.651.
C5.651.
C16.320.

Muscular Dystrophy, Emery-Dreifuss

C16.320.577.350

C5.651.
C10.668.
C16.320.

Muscular Dystrophy, Facioscapulohumeral

Muscular Dystrophy, Oculopharyngeal

Myotonic Dystrophy

C16.320.577.400
C16.320.577.450
C16.320.577.500

C5.651.
C5.651.
C5.651.
C10.574.
C10.668.
C10.668.
C16.320.

Myasthenic Syndromes, Congenital

Nail-Patella Syndrome

C16.320.590
C16.320.600

C10.668.
C5.550.
C16.131.
C17.800.

Neoplastic Syndromes, Hereditary

Adenomatous Polyposis Coli

C16.320.700
C16.320.700.100

C4.700
C4.557.
C4.588.
C6.301.
C6.405.
C6.405.

Gardner Syndrome

C16.320.700.100.393

C4.557.
C4.588.
C6.301.
C6.405.
C6.405.
C16.131.

Basal Cell Nevus Syndrome

C16.320.700.175

C4.182.
C4.557.
C5.116.
C5.500.

Colorectal Neoplasms, Hereditary Nonpolyposis

C16.320.700.250

C7.320.
C4.588.
C6.301.
C6.405.
C6.405.
C18.452.

Dysplastic Nevus Syndrome

Exostoses, Multiple Hereditary

C16.320.700.305
C16.320.700.330

C4.557.
C4.557.
C5.116.
C5.116.

Hamartoma Syndrome, Multiple

C16.320.700.435

C4.445.
C4.651.
C4.700.

Li-Fraumeni Syndrome

Multiple Endocrine Neoplasia

C16.320.700.600
C16.320.700.630

C4.700.
C18.452.
C4.588.
C4.651.
C4.700.
C19.344.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Neoplastic Syndromes, Hereditary

Multiple Endocrine Neoplasia

Multiple Endocrine Neoplasia Type 1

Multiple Endocrine Neoplasia Type 1

C16.320.700.630.500

C4.588.

C4.651.

Multiple Endocrine Neoplasia Type 2a

C16.320.700.630.505

C4.700.

C19.344.

Multiple Endocrine Neoplasia Type 2b

C16.320.700.630.510

C4.588.

C4.651.

Wilms Tumor

C16.320.700.642

C4.700.

C19.344.

C4.588.

C4.588.

C4.700.

C12.758.

C12.777.

C13.351.

Denys-Drash Syndrome

C16.320.700.642.220

C13.351.

C4.588.

C4.557.

C4.588.

C4.700.

C12.706.

C12.758.

C12.777.

C13.351.

C13.351.

C13.351.

C16.131.

WAGR Syndrome

C16.320.700.642.950

C13.351.

C16.131.

C4.557.

C4.588.

C4.700.

C10.597.

C11.250.

C11.270.

C11.941.

C12.706.

C12.758.

C12.777.

C13.351.

C13.351.

C13.351.

C16.131.

C16.131.

C16.131.

C16.320.

C16.320.

Neurofibromatoses

C16.320.700.645

C4.557.

C4.700.

C10.562.

C10.574.

C16.320.

Neurofibromatosis 1

C16.320.700.645.650

C4.557.

C4.700.

C10.562.

C10.574.

C10.668.

C16.320.

Neurofibromatosis 2

C16.320.700.645.655

C4.557.

C4.557.

C4.557.

C4.588.

C4.700.

C9.218.

C9.647.

C10.292.

C10.292.

C10.551.

C10.562.

C10.574.

C16.320.

Peutz-Jeghers Syndrome

C16.320.700.705

C4.700.

C6.405.

C17.800.

Osteogenesis Imperfecta

C16.320.737

C5.116.

C17.300.

Pain Insensitivity, Congenital

C16.320.775

C10.668.

Skin Diseases, Genetic

C16.320.850

C17.800.

Albinism

C16.320.850.80

C11.270.

C16.320.

C16.320.

C17.800.

Albinism, Ocular

C16.320.850.80.90

C17.800.

C18.452.

C17.800.

C18.452.

C11.270.

C16.320.

C16.320.

C17.800.

Albinism, Oculocutaneous

C16.320.850.80.100

C17.800.

C18.452.

C11.270.

C16.320.

C16.320.

C17.800.

Hermanski-Pudlak Syndrome

C16.320.850.80.100.400

C17.800.

C18.452.

C11.270.

C15.378.

C15.378.

C15.378.

C15.378.

C16.320.

C16.320.

C16.320.

C17.800.

C17.800.

Piebaldism

C16.320.850.80.600

C18.452.

C16.320.

C16.320.

C17.800.

C17.800.

Cutis Laxa

C16.320.850.180

C18.452.

C17.300.

C17.800.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Skin Diseases, Genetic

Dermatitis, Atopic

Dermatitis, Atopic	C16.320.850.210	C17.800. C17.800.	C17.800. C20.543.
Dyskeratosis Congenita	C16.320.850.235	C16.131. C17.800.	C16.320. C17.800.
Ectodermal Dysplasia	C16.320.850.250	C16.131. C17.800.	C16.131. C17.800.
Ectodermal Dysplasia 1, Anhidrotic	C16.320.850.250.198	C16.131. C16.320. C17.800.	C16.131. C17.800.
Ectodermal Dysplasia 3, Anhidrotic	C16.320.850.250.298	C16.131. C17.800.	C16.131.
Ectodermal Dysplasia, Hypohidrotic, Autosomal Recessive	C16.320.850.250.348	C16.131. C17.800.	C16.131. C17.800.
Ellis-Van Creveld Syndrome	C16.320.850.250.398	C5.116. C16.131. C17.800.	C16.131. C17.800.
Focal Dermal Hypoplasia	C16.320.850.250.424	C5.116. C16.131. C17.800.	C16.131. C16.320. C17.800.
Neurocutaneous Syndromes	C16.320.850.250.712	C10.562 C16.131. C17.800.	C16.131. C17.800.
Pachyonychia Congenita	C16.320.850.250.856	C16.131. C17.800.	C17.800. C17.800.
Ehlers-Danlos Syndrome	C16.320.850.260	C14.907. C16.131. C17.800.	C15.378. C17.300. C17.800.
Epidermolysis Bullosa	C16.320.850.275	C16.131. C17.800.	C17.800. C17.800.
Epidermolysis Bullosa Dystrophica	C16.320.850.275.160	C16.131. C17.800.	C17.300. C17.800.
Epidermolysis Bullosa, Junctional	C16.320.850.275.170	C16.131. C17.800.	C17.800. C17.800.
Epidermolysis Bullosa Simplex	C16.320.850.275.180	C16.131. C17.800.	C17.800. C17.800.
Ichthyosiform Erythroderma, Congenital	C16.320.850.400	C16.131. C17.800. C17.800.	C16.614. C17.800.
Hyperkeratosis, Epidermolytic	C16.320.850.400.375	C16.131. C17.800.	C16.614. C17.800.
Ichthyosis, Lamellar	C16.320.850.400.410	C16.131. C17.800. C17.800.	C16.614. C17.800.
Ichthyosis Bullosa of Siemens	C16.320.850.402	C16.131. C17.800.	C16.614. C17.800.
Ichthyosis Vulgaris	C16.320.850.405	C16.131. C17.800.	C17.800. C17.800.
Ichthyosis, X-Linked	C16.320.850.408	C16.131. C16.320. C17.800. C17.800.	C16.320. C16.614. C17.800. C18.452.
Incontinentia Pigmenti	C16.320.850.420	C16.131. C17.800. C17.800.	C16.131. C17.800.
Keratoderma, Palmoplantar	C16.320.850.475	C17.800.	C17.800.
Keratoderma, Palmoplantar, Diffuse	C16.320.850.475.440	C17.800.	C17.800.
Keratoderma, Palmoplantar, Epidermolytic	C16.320.850.475.440.500	C17.800.	C17.800.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Skin Diseases, Genetic

Keratoderma, Palmoplantar

Papillon-Lefevre Disease

Papillon-Lefevre Disease

Keratosis Follicularis

Leukokeratosis, Hereditary Mucosal

Lipoid Proteinosis of Urbach and Wiethe

Pemphigus, Benign Familial

Porokeratosis

Porphyria, Erythropoietic

Porphyrias, Hepatic

Coproporphyrin, Hereditary

Porphyria, Acute Intermittent

Porphyria Cutanea Tarda

Porphyria, Hepatoerythropoietic

Porphyria, Variegate

Protoporphyrin, Erythropoietic

Pseudoxanthoma Elasticum

Rothmund-Thomson Syndrome

Sjogren-Larsson Syndrome

Trichothiodystrophy Syndromes

Xeroderma Pigmentosum

Werner Syndrome

Infant, Newborn, Diseases

Amniotic Band Syndrome

Anemia, Neonatal

Fetofetal Transfusion

Fetomaternal Transfusion

C16.320.850.475.600	C17.800.	C17.800.
C16.320.850.490	C17.800.	C17.800.
C16.320.850.542	C17.800.	
C16.320.850.595	C8.618.	C17.800.
C16.320.850.700	C17.800.	C17.800.
	C20.111.	
C16.320.850.730	C17.800.	C17.800.
C16.320.850.738	C16.320.	C17.800.
	C17.800.	C18.452.
	C18.452.	C18.452.
C16.320.850.742	C6.552.	C16.320.
	C17.800.	C17.800.
	C18.452.	C18.452.
	C18.452.	C18.452.
C16.320.850.742.74	C6.552.	C16.320.
	C17.800.	C17.800.
	C18.452.	C18.452.
	C18.452.	C18.452.
C16.320.850.742.150	C6.552.	C16.320.
	C17.800.	C17.800.
	C18.452.	C18.452.
	C18.452.	C18.452.
C16.320.850.742.250	C6.552.	C16.320.
	C17.800.	C17.800.
	C18.452.	C18.452.
	C18.452.	C18.452.
C16.320.850.742.437	C6.552.	C16.320.
	C17.800.	C17.800.
	C18.452.	C18.452.
	C18.452.	C18.452.
C16.320.850.742.625	C6.552.	C16.320.
	C17.800.	C17.800.
	C18.452.	C18.452.
	C18.452.	C18.452.
C16.320.850.742.812	C6.552.	C16.320.
	C17.800.	C17.800.
	C18.452.	C18.452.
	C18.452.	C18.452.
C16.320.850.750	C14.907.	C15.378.
	C16.131.	C17.300.
	C17.800.	C17.800.
C16.320.850.765	C16.131.	C16.614.
	C17.800.	C17.800.
	C18.452.	C18.452.
C16.320.850.820	C16.131.	C16.320.
	C16.614.	C17.800.
	C17.800.	C17.800.
	C18.452.	C18.452.
C16.320.850.895	C16.131.	C16.131.
	C17.800.	C17.800.
C16.320.850.970	C4.834.	C16.131.
	C17.800.	C17.800.
	C17.800.	C17.800.
	C18.452.	C18.452.
C16.320.925	C18.452.	
C16.614		
C16.614.42		
C16.614.53	C15.378.	
C16.614.53.344	C15.378.	
C16.614.53.511	C15.378.	

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Infant, Newborn, Diseases
Asphyxia Neonatorum

Asphyxia Neonatorum	C16.614.92		
Birth Injuries	C16.614.131	C21.866.	
Paralysis, Obstetric	C16.614.131.587	C21.866.	
Cystic Fibrosis	C16.614.213	C6.689.	C8.381.
		C16.320.	
Epilepsy, Benign Neonatal	C16.614.258	C10.228.	
Erythroblastosis, Fetal	C16.614.304	C13.703.	C15.378.
		C16.300.	C20.188.
Kernicterus	C16.614.304.502	C10.228.	C15.378.
		C18.452.	C20.188.
		C23.550.	
Hemorrhagic Disease of Newborn	C16.614.378	C15.378.	C15.378.
		C18.654.	C18.654.
Hernia, Umbilical	C16.614.390	C23.300.	
Hydrocephalus	C16.614.414	C10.228.	C10.228.
Dandy-Walker Syndrome	C16.614.414.200	C10.228.	C10.228.
		C10.228.	C10.500.
		C16.131.	
Hydrophthalmos	C16.614.438	C11.250.	C11.525.
		C16.131.	
Hyperbilirubinemia, Neonatal	C16.614.451	C23.550.	
Jaundice, Neonatal	C16.614.451.500	C23.550.	
Jaundice, Chronic Idiopathic	C16.614.451.500.250	C16.320.	C18.452.
Hyperostosis, Cortical, Congenital	C16.614.465	C5.116.	C5.116.
Ichthyosis	C16.614.492	C16.131.	C17.800.
		C17.800.	
Ichthyosiform Erythroderma, Congenital	C16.614.492.400	C16.131.	C16.320.
		C17.800.	C17.800.
		C17.800.	
Hyperkeratosis, Epidermolytic	C16.614.492.400.375	C16.131.	C16.320.
		C17.800.	C17.800.
		C17.800.	
Ichthyosis, Lamellar	C16.614.492.400.410	C16.131.	C16.320.
		C17.800.	C17.800.
		C17.800.	
Ichthyosis Bullosa of Siemens	C16.614.492.410	C16.131.	C16.320.
		C17.800.	C17.800.
		C17.800.	
Ichthyosis, X-Linked	C16.614.492.420	C16.131.	C16.320.
		C16.320.	C16.320.
		C17.800.	C17.800.
		C17.800.	C18.452.
Sjogren-Larsson Syndrome	C16.614.492.723	C16.131.	C16.320.
		C16.320.	C17.800.
		C17.800.	C17.800.
		C18.452.	C18.452.
Infant, Premature, Diseases	C16.614.521		
Bronchopulmonary Dysplasia	C16.614.521.125	C8.381.	
Leukomalacia, Periventricular	C16.614.521.450	C10.228.	C10.228.
		C14.907.	
Respiratory Distress Syndrome, Newborn	C16.614.521.563	C8.381.	C8.618.
Hyaline Membrane Disease	C16.614.521.563.475	C8.381.	C8.618.
Retinopathy of Prematurity	C16.614.521.731	C11.768.	
Meconium Aspiration Syndrome	C16.614.580	C8.381.	C8.618.
		C13.703.	C16.300.
Mobius Syndrome	C16.614.595	C10.292.	C16.131.
Neonatal Abstinence Syndrome	C16.614.610	C21.739.	F3.900.
Nystagmus, Congenital	C16.614.643	C10.292.	C11.590.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Infant, Newborn, Diseases
Ophthalmia Neonatorum

Ophthalmia Neonatorum	C16.614.677	C1.252. C1.539. C11.294.	C1.252. C11.187.
Persistent Fetal Circulation Syndrome	C16.614.694	C8.381.	
Persistent Hyperinsulinemia Hypoglycemia of Infancy	C16.614.716	C18.452.	C18.452.
Rothmund-Thomson Syndrome	C16.614.760	C16.131. C17.800. C18.452.	C16.320. C17.800.
Sclerema Neonatorum	C16.614.810	C16.131.	C17.800.
Severe Combined Immunodeficiency	C16.614.815	C18.452.	C20.673.
X-Linked Combined Immunodeficiency Diseases	C16.614.815.500	C16.320.	C20.673.
Syphilis, Congenital	C16.614.868	C1.252.	C1.252.
Thanatophoric Dysplasia	C16.614.890	C5.116. C5.660.	C5.116. C16.131.
Thrombocytopenia, Neonatal Alloimmune	C16.614.899	C15.378.	
Toxoplasmosis, Congenital	C16.614.909	C3.752.	C10.228.
Wolman Disease	C16.614.947	C16.320. C18.452. C18.452.	C16.320. C18.452.