MeSH Tree Structures - 2012

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		
22q11 Deletion Syndrome	C16.131.77.19	C5.660.	C14.240.
		C14.280.	C15.604.
		C16.131.	C16.131.
		C16.131.	C16.131.
		C16.320.	C19.642.
DiGeorge Syndrome	C16.131.77.19.500	C5.660.	C14.240.
		C14.280.	C15.604.
		C16.131.	C16.131.
		C16.131.	C16.131.
		C16.320.	C19.642.
Alagille Syndrome	C16.131.77.65	C6.130.	C6.552.
		C14.240.	C16.131.
		C16.320.	
Alstrom Syndrome	C16.131.77.80	C10.500.	C10.574.
		C10.668.	C11.270.
		C16.131.	C16.320.
		C16.320.	
Angelman Syndrome	C16.131.77.95	C10.228.	C16.131.
		C16.320.	
Bardet-Biedl Syndrome	C16.131.77.112	C10.228.	
Barth Syndrome	C16.131.77.121	C14.240.	C14.280.
		C16.131.	C16.320.
		C16.320.	C18.452.
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.
Carney Complex	C16.131.77.229	C4.557.	C4.588.
		C14.280.	C16.131.
CHARGE Syndrome	C16.131.77.239	C16.320.	
Cockayne Syndrome	C16.131.77.250	C5.116.	C10.574.
		C16.320.	C16.320.
		C18.452.	
Costello Syndrome	C16.131.77.256	C5.660.	C16.320.
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.	C11.768.
		C11.966.	C16.320.
		C23.888.	

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Congenital Abnormalities
Abnormalities, Multiple
Deaf-Blind Disorders
Wolfram Syndrome

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.
Donohue Syndrome	C16.131.77.313	C5.660.	C16.320.
		C18.452.	C19.246.
Down Syndrome	C16.131.77.327	C10.597.	C16.131.
		C16.320.	
Ectodermal Dysplasia	C16.131.77.350	C16.131.	C16.320.
		C17.800.	C17.800.
Ectodermal Dysplasia 1, Anhidrotic	C16.131.77.350.198	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.	
Fraser Syndrome	C16.131.77.371	C11.250.	C16.131.
Gardner Syndrome	C16.131.77.393	C4.557.	C4.588.
		C4.588.	C4.700.
		C6.301.	C6.405.
		C6.405.	C6.405.
		C6.405.	C16.320.
Heterotaxy Syndrome	C16.131.77.401	C14.240.	C14.280.
		C15.604.	C16.131.
Holoprosencephaly	C16.131.77.410	C5.660.	C10.500.
		C16.131.	C16.131.
		C16.131.	C16.131.
		C16.320.	
Incontinentia Pigmenti	C16.131.77.445	C16.131.	C16.320.
		C17.800.	C17.800.
		C17.800.	
Isolated Noncompaction of the Ventricular Myocardium	C16.131.77.477	C14.240.	C14.280.
		C14.280.	C16.131.
		C16.320.	
Laurence-Moon Syndrome	C16.131.77.509	C10.228.	
LEOPARD Syndrome	C16.131.77.525	C5.660.	C14.240.
		C14.280.	C14.280.
		C16.131.	C16.131.
		C17.800.	
Loeys-Dietz Syndrome	C16.131.77.537	C5.660.	C14.907.
		C14.907.	C14.907.
		C16.320.	
Marfan Syndrome	C16.131.77.550	C5.116.	C14.240.
		C14.280.	C16.131.
		C16.320.	C17.300.
Mobius Syndrome	C16.131.77.578	C10.292.	C16.614.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Congenital Abnormalities
Abnormalities, Multiple
Monilethrix

Monilethrix	C16.131.77.592	C16.320.	C17.800.
		C17.800.	
Nail-Patella Syndrome	C16.131.77.606	C5.550.	C16.320.
		C17.800.	
Netherton Syndrome	C16.131.77.619	C16.131.	C16.320.
		C16.320.	C16.614.
		C17.800.	C17.800.
		C17.800.	C17.800.
Nevus, Sebaceous of Jadassohn	C16.131.77.633	C4.557.	C10.562.
Oculocerebrorenal Syndrome	C16.131.77.661	C10.228.	C12.777.
		C13.351.	C16.320.
		C16.320.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Orofaciodigital Syndromes	C16.131.77.677	C5.116.	C5.660.
		C16.131.	C16.131.
		C16.320.	
Pallister-Hall Syndrome	C16.131.77.690	C4.445.	C4.588.
		C5.660.	C10.228.
		C10.228.	C10.551.
		C16.131.	
Pentalogy of Cantrell	C16.131.77.696	C10.500.	C16.131.
POEMS Syndrome	C16.131.77.703	C10.668.	C15.378.
		C20.683.	
Prader-Willi Syndrome	C16.131.77.730	C10.597.	C16.131.
		C16.320.	C18.654.
Prolidase Deficiency	C16.131.77.735	C16.131.	C16.320.
		C16.320.	
Proteus Syndrome	C16.131.77.740	C4.445.	C4.651.
		C5.116.	C5.660.
		C16.131.	
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.	C5.660.
		C10.597.	C16.131.
		C16.131.	C16.320.
Short Rib-Polydactyly Syndrome	C16.131.77.850	C5.116.	C5.660.
		C16.131.	
Silver-Russell Syndrome	C16.131.77.855	C5.660.	C16.131.
		C16.320.	C16.320.
Smith-Lemli-Opitz Syndrome	C16.131.77.860	C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Smith-Magenis Syndrome	C16.131.77.879	C10.281.	C16.131.
		C16.320.	
Sotos Syndrome	C16.131.77.889	C16.131.	C16.320.
Trichothiodystrophy Syndromes	C16.131.77.899	C16.131.	C16.320.
		C17.800.	C17.800.
Waardenburg's Syndrome	C16.131.77.938		
Weill-Marchesani Syndrome	C16.131.77.941	C5.116.	C5.116.
		C11.270.	C16.320.
		C16.320.	C17.300.
Wolf-Hirschhorn Syndrome	C16.131.77.944	C16.131.	C16.320.

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.
		C19.246.	C19.700.
Zellweger Syndrome	C16.131.77.970	C6.552.	C10.228.
		C12.777.	C13.351.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Abnormalities, Radiation-Induced	C16.131.80	C26.733.	N6.850.
Abnormalities, Severe Teratoid	C16.131.85		
Anencephaly	C16.131.85.197	C10.500.	C16.131.
Twins, Conjoined	C16.131.85.806		
Aicardi Syndrome	C16.131.162	C10.500.	C11.270.
		C16.131.	C16.320.
		C16.320.	
Cardiovascular Abnormalities	C16.131.240	C14.240	
Heart Defects, Congenital	C16.131.240.400	C14.240.	C14.280.
22q11 Deletion Syndrome	C16.131.240.400.21	C5.660.	C14.240.
		C14.280.	C15.604.
		C16.131.	C16.131.
		C16.131.	C16.131.
		C16.320.	C19.642.
DiGeorge Syndrome	C16.131.240.400.21.500	C5.660.	C14.240.
		C14.280.	C15.604.
		C16.131.	C16.131.
		C16.131.	C16.131.
		C16.320.	C19.642.
Alagille Syndrome	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.	
Barth Syndrome	C16.131.240.400.172	C14.240.	C14.280.
		C16.131.	C16.320.
		C16.320.	C18.452.
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.
		C16.131.	
Kartagener Syndrome	C16.131.240.400.280.500	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.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities			
Congenital Abnormalities			
Cardiovascular Abnormalities			
Heart Defects, Congenital			
Heart Septal Defects			
Heart Septal Defects, Atrial			
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.
Heterotaxy Syndrome	C16.131.240.400.592	C14.240.	C14.280.
		C15.604.	C16.131.
Hypoplastic Left Heart Syndrome	C16.131.240.400.625	C14.240.	C14.280.
Isolated Noncompaction of the Ventricular Myocardium	C16.131.240.400.655	C14.240.	C14.280.
		C14.280.	C16.131.
		C16.320.	
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.
		C16.131.	
Long QT Syndrome	C16.131.240.400.715	C14.280.	C23.550.
Andersen Syndrome	C16.131.240.400.715.70	C14.280.	C23.550.
Jervell-Lange Nielsen Syndrome	C16.131.240.400.715.440	C14.280.	
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.
		C16.320.	C17.300.
Noonan Syndrome	C16.131.240.400.784	C5.660.	C14.240.
		C14.280.	C16.131.
		C17.300.	
Tetralogy of Fallot	C16.131.240.400.849	C14.240.	C14.280.
Transposition of Great Vessels	C16.131.240.400.915	C14.240.	C14.280.
Double Outlet Right Ventricle	C16.131.240.400.915.300	C14.240.	C14.280.
Tricuspid Atresia	C16.131.240.400.920	C14.240.	C14.280.
		C14.280.	
Trilogy of Fallot	C16.131.240.400.960	C14.240.	C14.280.
Turner Syndrome	C16.131.240.400.970	C12.706.	C12.706.
		C13.351.	C13.351.
		C14.240.	C14.280.
		C16.131.	C16.131.
		C16.131.	C16.320.
		C19.391.	C19.391.
Wolff-Parkinson-White Syndrome	C16.131.240.400.980	C14.280.	
Vascular Malformations	C16.131.240.850	C14.240.	
Arterio-Arterial Fistula	C16.131.240.850.500	C14.240.	C14.907.
		C23.300.	
Arteriovenous Malformations	C16.131.240.850.750	C14.240.	C14.907.
Arteriovenous Fistula	C16.131.240.850.750.125	C14.240.	C14.240.
		C14.907.	C14.907.
		C23.300.	
Intracranial Arteriovenous Malformations	C16.131.240.850.750.295	C10.228.	C10.500.
		C14.240.	C14.240.
		C14.907.	C14.907.
		C16.131.	C16.131.
Vein of Galen Malformations	C16.131.240.850.750.295.500	C10.228.	C10.500.
		C14.240.	C14.240.
		C14.907.	C14.907.
		C16.131.	C16.131.
Central Nervous System Vascular Malformations	C16.131.240.850.875	C10.500.	C14.240.
		C16.131.	
Intracranial Arteriovenous Malformations	C16.131.240.850.875.500	C10.228.	C10.500.
		C14.240.	C14.240.
		C14.907.	C14.907.
		C16.131.	C16.131.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities			
Congenital Abnormalities			
Cardiovascular Abnormalities			
Vascular Malformations			
Central Nervous System Vascular Malformations			
Intracranial Arteriovenous Malformations			
Vein of Galen Malformations	C16.131.240.850.875.500.500	C10.228.	C10.500.
		C14.240.	C14.240.
		C14.907.	C14.907.
		C16.131.	C16.131.
Pulmonary Atresia	C16.131.240.850.906	C14.240.	C14.280.
Scimitar Syndrome	C16.131.240.850.937	C8.381.	C8.695.
		C14.240.	C14.907.
		C16.131.	
Single Umbilical Artery	C16.131.240.850.952	C14.240.	
Telangiectasia, Hereditary Hemorrhagic	C16.131.240.850.968	C14.907.	C14.907.
		C15.378.	
Chromosome Disorders	C16.131.260	C16.320.	
22q11 Deletion Syndrome	C16.131.260.19	C5.660.	C14.240.
		C14.280.	C15.604.
		C16.131.	C16.131.
		C16.131.	C16.131.
		C16.320.	C19.642.
DiGeorge Syndrome	C16.131.260.19.500	C5.660.	C14.240.
		C14.280.	C15.604.
		C16.131.	C16.131.
		C16.131.	C16.131.
		C16.320.	C19.642.
Angelman Syndrome	C16.131.260.40	C10.228.	C16.131.
		C16.320.	
Beckwith-Wiedemann Syndrome	C16.131.260.80	C16.131.	C16.320.
Branchio-Oto-Renal Syndrome	C16.131.260.90	C16.131.	C16.320.
Cri-du-Chat Syndrome	C16.131.260.190	C10.597.	C16.131.
		C16.320.	
De Lange Syndrome	C16.131.260.210	C10.597.	C16.131.
		C16.320.	
Down Syndrome	C16.131.260.260	C10.597.	C16.131.
		C16.320.	
Holoprosencephaly	C16.131.260.380	C5.660.	C10.500.
		C16.131.	C16.131.
		C16.131.	C16.131.
		C16.320.	
Jacobsen Distal 11q Deletion Syndrome	C16.131.260.440	C15.378.	C16.320.
Prader-Willi Syndrome	C16.131.260.700	C10.597.	C16.131.
		C16.320.	C18.654.
Rubinstein-Taybi Syndrome	C16.131.260.790	C5.116.	C5.660.
		C10.597.	C16.131.
		C16.131.	C16.320.
Sex Chromosome Disorders	C16.131.260.830	C16.320.	
Fragile X Syndrome	C16.131.260.830.300	C10.597.	C16.320.
		C16.320.	C16.320.
Orofaciodigital Syndromes	C16.131.260.830.670	C5.116.	C5.660.
		C16.131.	C16.131.
		C16.320.	
Sex Chromosome Disorders of Sex Development	C16.131.260.830.835	C12.706.	C13.351.
		C16.131.	C16.320.
		C19.391.	
Gonadal Dysgenesis, Mixed	C16.131.260.830.835.249	C12.706.	C12.706.
		C13.351.	C13.351.
		C16.131.	C16.131.
		C16.320.	C19.391.
		C19.391.	
Klinefelter Syndrome	C16.131.260.830.835.500	C12.706.	C13.351.
		C16.131.	C16.320.
		C19.391.	C19.391.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities			
Congenital Abnormalities			
Chromosome Disorders			
Sex Chromosome Disorders			
Sex Chromosome Disorders of Sex Development			
Turner Syndrome			
Turner Syndrome	C16.131.260.830.835.750	C12.706.	C12.706.
		C13.351.	C13.351.
		C14.240.	C14.280.
		C16.131.	C16.131.
		C16.131.	C16.320.
		C19.391.	C19.391.
Silver-Russell Syndrome	C16.131.260.870	C5.660.	C16.131.
		C16.320.	C16.320.
Smith-Magenis Syndrome	C16.131.260.887	C10.281.	C16.131.
		C16.320.	
Sotos Syndrome	C16.131.260.905	C16.131.	C16.320.
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.
		C19.391.	
Williams Syndrome	C16.131.260.970	C10.597.	C14.280.
		C16.320.	
Wolf-Hirschhorn Syndrome	C16.131.260.985	C16.131.	C16.320.
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.
		C19.391.	
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.
Fraser Syndrome	C16.131.384.442	C11.250.	C16.131.
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.	
Retinal Dysplasia	C16.131.384.784	C11.250.	C11.270.
		C11.768.	C16.320.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Congenital Abnormalities
Lymphatic Abnormalities

Lymphatic Abnormalities	C16.131.482	C15.604.	
22q11 Deletion Syndrome	C16.131.482.249	C5.660.	C14.240.
		C14.280.	C15.604.
		C16.131.	C16.131.
		C16.131.	C16.131.
		C16.320.	C19.642.
DiGeorge Syndrome	C16.131.482.249.500	C5.660.	C14.240.
		C14.280.	C15.604.
		C16.131.	C16.131.
		C16.131.	C16.131.
		C16.320.	C19.642.
Lymphangiectasis, Intestinal	C16.131.482.500	C15.604.	C15.604.
Musculoskeletal Abnormalities	C16.131.621	C5.660	
Arthrogryposis	C16.131.621.77	C5.550.	C5.651.
		C5.660.	
Campomelic Dysplasia	C16.131.621.142	C5.660.	
Cervical Rib Syndrome	C16.131.621.174	C10.668.	C14.907.
Craniofacial Abnormalities	C16.131.621.207	C5.660.	
22q11 Deletion Syndrome	C16.131.621.207.103	C5.660.	C14.240.
		C14.280.	C15.604.
		C16.131.	C16.131.
		C16.131.	C16.131.
		C16.320.	C19.642.
DiGeorge Syndrome	C16.131.621.207.103.500	C5.660.	C14.240.
		C14.280.	C15.604.
		C16.131.	C16.131.
		C16.131.	C16.131.
		C16.320.	C19.642.
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.	C5.660.
		C16.131.	C16.131.
Acrocephalosyndactylia	C16.131.621.207.240.100	C5.116.	C5.116.
		C5.660.	C5.660.
		C5.660.	C5.660.
		C5.660.	C16.131.
		C16.131.	C16.131.
		C16.131.	
Holoprosencephaly	C16.131.621.207.410	C5.660.	C10.500.
		C16.131.	C16.131.
		C16.131.	C16.131.
		C16.320.	
LEOPARD Syndrome	C16.131.621.207.525	C5.660.	C14.240.
		C14.280.	C14.280.
		C16.131.	C16.131.
		C17.800.	
Macrocephaly	C16.131.621.207.532	C5.660.	C10.500.
		C16.131.	
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.	

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities			
Congenital Abnormalities			
Musculoskeletal Abnormalities			
Craniofacial Abnormalities			
Maxillofacial Abnormalities			
Jaw Abnormalities			
Cleft Palate	C16.131.621.207.540.460.185	C5.500. C7.320. C7.650. C16.131.	C5.660. C7.465. C7.650. C16.131.
Micrognathism	C16.131.621.207.540.460.457	C5.500. C7.320. C16.131.	C5.660. C7.650.
Pierre Robin Syndrome	C16.131.621.207.540.460.606	C5.500. C7.320. C16.131.	C5.660. C7.650.
Prognathism	C16.131.621.207.540.460.655	C5.500. C5.660. C7.320. C16.131.	C5.500. C7.320. C7.650.
Retrognathism	C16.131.621.207.540.460.813	C5.500. C5.660. C7.320. C16.131.	C5.500. C7.320. C7.650.
Microcephaly	C16.131.621.207.620	C5.660. C16.131.	C10.500.
Noonan Syndrome	C16.131.621.207.690	C5.660. C14.280. C17.300.	C14.240. C16.131.
Orofaciodigital Syndromes	C16.131.621.207.700	C5.116. C16.131. C16.320.	C5.660. C16.131.
Plagiocephaly	C16.131.621.207.707	C5.660.	
Craniosynostoses	C16.131.621.207.707.249	C5.116. C5.660. C16.131.	C5.660. C5.660. C16.131.
Acrocephalosyndactylia	C16.131.621.207.707.249.100	C5.116. C5.660. C5.660. C5.660. C16.131. C16.131.	C5.116. C5.660. C5.660. C16.131. C16.131.
Plagiocephaly, Nonsynostotic	C16.131.621.207.707.624	C5.660.	
Platybasia	C16.131.621.207.720	C5.116. C5.660.	C5.116.
Rubinstein-Taybi Syndrome	C16.131.621.207.850	C5.116. C10.597. C16.131.	C5.660. C16.131. C16.320.
Funnel Chest	C16.131.621.386	C5.116.	C5.660.
Gastroschisis	C16.131.621.417	C5.660.	C23.300.
Hajdu-Cheney Syndrome	C16.131.621.445	C5.116. C16.320.	C5.116.
Hip Dislocation, Congenital	C16.131.621.449	C5.550.	C5.660.
Klippel-Feil Syndrome	C16.131.621.551	C5.116.	C5.660.
Laryngomalacia	C16.131.621.568	C5.182. C9.400.	C8.360. C17.300.
Limb Deformities, Congenital	C16.131.621.585	C5.660.	
Arachnodactyly	C16.131.621.585.174	C5.660.	
Brachydactyly	C16.131.621.585.262	C5.660.	
Ectromelia	C16.131.621.585.350	C5.660.	
Foot Deformities, Congenital	C16.131.621.585.380	C5.330.	C5.660.
Clubfoot	C16.131.621.585.380.500	C5.330.	C5.660.
Hand Deformities, Congenital	C16.131.621.585.425	C5.390.	C5.660.
Lower Extremity Deformities, Congenital	C16.131.621.585.512	C5.660.	
Polydactyly	C16.131.621.585.600	C5.660.	

Congenital, Hereditary, and Neonatal Diseases and Abnormalities			
Congenital Abnormalities			
Musculoskeletal Abnormalities			
Limb Deformities, Congenital			
Polydactyly			
Pallister-Hall Syndrome			
Pallister-Hall Syndrome	C16.131.621.585.600.374	C4.445. C5.660. C10.228. C16.131.	C4.588. C10.228. C10.551.
Short Rib-Polydactyly Syndrome	C16.131.621.585.600.750	C5.116. C16.131.	C5.660.
Proteus Syndrome	C16.131.621.585.620	C4.445. C5.116. C16.131.	C4.651. C5.660.
Syndactyly	C16.131.621.585.800	C5.116. C5.660.	C5.660. C16.131.
Acrocephalosyndactylia	C16.131.621.585.800.100	C5.116. C5.660. C5.660. C5.660. C16.131. C16.131.	C5.116. C5.660. C5.660. C16.131. C16.131. C16.131.
Poland Syndrome	C16.131.621.585.800.756	C5.116. C5.660.	C5.660. C16.131.
Thanatophoric Dysplasia	C16.131.621.585.984	C5.116. C5.660. C16.614.	C5.116. C16.320.
Upper Extremity Deformities, Congenital	C16.131.621.585.988	C5.660.	
Synostosis	C16.131.621.906	C5.116.	C5.660.
Antley-Bixler Syndrome Phenotype	C16.131.621.906.181	C5.116. C16.320.	C5.660. C18.452.
Craniosynostoses	C16.131.621.906.364	C5.116. C5.660. C16.131.	C5.660. C5.660. C16.131.
Acrocephalosyndactylia	C16.131.621.906.364.100	C5.116. C5.660. C5.660. C5.660. C16.131. C16.131.	C5.116. C5.660. C5.660. C16.131. C16.131. C16.131.
Syndactyly	C16.131.621.906.819	C5.116. C5.660.	C5.660. C16.131.
Acrocephalosyndactylia	C16.131.621.906.819.100	C5.116. C5.660. C5.660. C5.660. C16.131. C16.131.	C5.116. C5.660. C5.660. C16.131. C16.131. C16.131.
Poland Syndrome	C16.131.621.906.819.756	C5.116. C5.660.	C5.660. C16.131.
Tracheobronchomalacia	C16.131.621.953	C5.182. C8.907.	C8.127. C17.300.
Bronchomalacia	C16.131.621.953.249	C5.182. C17.300.	C8.127.
Tracheomalacia	C16.131.621.953.500	C5.182. C17.300.	C8.907.
Nervous System Malformations	C16.131.666	C10.500	
Agenesis of Corpus Callosum	C16.131.666.34	C10.500.	C23.300.
Acrocallosal Syndrome	C16.131.666.34.500	C10.500.	
Aicardi Syndrome	C16.131.666.34.687	C10.500. C16.131. C16.320.	C11.270. C16.320.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Congenital Abnormalities
Nervous System Malformations
Agenesis of Corpus Callosum
Holoprosencephaly

Holoprosencephaly	C16.131.666.34.875	C5.660. C16.131. C16.131. C16.320.	C10.500. C16.131. C16.131.
Central Nervous System Cysts	C16.131.666.142	C4.588. C10.551.	C10.500.
Arachnoid Cysts	C16.131.666.142.100	C4.182. C10.500.	C4.588. C10.551.
Colloid Cysts	C16.131.666.142.200	C4.182. C10.500.	C4.588. C10.551.
Central Nervous System Vascular Malformations	C16.131.666.190	C10.500. C16.131.	C14.240.
Central Nervous System Venous Angioma	C16.131.666.190.100	C4.557. C14.240.	C10.500.
Hemangioma, Cavernous, Central Nervous System	C16.131.666.190.200	C4.557. C14.240. C15.378.	C10.500. C14.907.
Intracranial Arteriovenous Malformations	C16.131.666.190.500	C10.228. C14.240. C14.907. C16.131.	C10.500. C14.240. C14.907. C16.131.
Vein of Galen Malformations	C16.131.666.190.500.500	C10.228. C14.240. C14.907. C16.131.	C10.500. C14.240. C14.907. 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.
Hereditary Sensory and Motor Neuropathy	C16.131.666.300	C10.500. C10.668.	C10.574. C16.320.
Alstrom Syndrome	C16.131.666.300.99	C10.500. C10.668. C16.131. C16.320.	C10.574. C11.270. C16.320.
Charcot-Marie-Tooth Disease	C16.131.666.300.200	C10.500. C10.668.	C10.574. C16.320.
Giant Axonal Neuropathy	C16.131.666.300.490	C10.500. C10.668. C16.320.	C10.574. C10.668.
Refsum Disease	C16.131.666.300.780	C10.228. C10.574. C16.320. C16.320. C18.452.	C10.500. C10.668. C16.320. C18.452. C18.452.
Spastic Paraplegia, Hereditary	C16.131.666.300.820	C10.500. C10.668.	C10.574. C16.320.
Hereditary Sensory and Autonomic Neuropathies	C16.131.666.310	C10.114. C10.500. C10.668. C16.320.	C10.314. C10.574. C10.668. C20.111.
Dysautonomia, Familial	C16.131.666.310.309	C10.114. C10.314. C10.574. C10.668. C20.111.	C10.177. C10.500. C10.668. C16.320.
Holoprosencephaly	C16.131.666.410	C5.660. C16.131. C16.131. C16.320.	C10.500. C16.131. C16.131.
Hydranencephaly	C16.131.666.450	C10.500.	

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Congenital Abnormalities
Nervous System Malformations
Malformations of Cortical Development

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. C16.131.	C10.500. C16.320.
Cobblestone Lissencephaly	C16.131.666.507.186.249	C10.500. C16.131.	C10.500.
Walker-Warburg Syndrome	C16.131.666.507.186.249.500	C10.500. C16.320.	C11.270.
Macrocephaly	C16.131.666.507.343	C5.660. C16.131.	C10.500.
Microcephaly	C16.131.666.507.500	C5.660. C16.131.	C10.500.
Neuronal Migration Disorders	C16.131.666.507.812	C10.500.	
Classical Lissencephalies and Subcortical Band Heterotopias	C16.131.666.507.812.230	C10.500. C16.131.	C10.500. C16.320.
Cobblestone Lissencephaly	C16.131.666.507.812.249	C10.500. C16.131.	C10.500.
Periventricular Nodular Heterotopia	C16.131.666.507.812.750	C10.500.	
Tuberous Sclerosis	C16.131.666.507.875	C4.445. C4.700. C10.562. C16.320.	C4.651. C10.500. C10.574. C16.320.
Neural Tube Defects	C16.131.666.680	C10.500.	
Anencephaly	C16.131.666.680.196	C10.500.	C16.131.
Arnold-Chiari Malformation	C16.131.666.680.291	C10.500.	
Encephalocele	C16.131.666.680.488	C10.500.	C23.300.
Meningocele	C16.131.666.680.598	C10.500.	C23.300.
Meningomyelocele	C16.131.666.680.610	C10.500.	
Pentalogy of Cantrell	C16.131.666.680.705	C10.500.	C16.131.
Spinal Dysraphism	C16.131.666.680.800	C10.500.	
Spina Bifida Cystica	C16.131.666.680.800.730	C10.500.	
Spina Bifida Occulta	C16.131.666.680.800.750	C10.500.	
Septo-Optic Dysplasia	C16.131.666.845	C10.500.	
Respiratory System Abnormalities	C16.131.740	C8.695	
Bronchogenic Cyst	C16.131.740.195	C4.182. C8.695.	C8.127.
Bronchopulmonary Sequestration	C16.131.740.214	C8.695.	
Choanal Atresia	C16.131.740.271	C8.460. C9.603.	C8.695.
Cystic Adenomatoid Malformation of Lung, Congenital	C16.131.740.290	C8.381.	C8.695.
Kartagener Syndrome	C16.131.740.501	C8.127. C8.695. C14.240. C16.131. C16.320.	C8.200. C9.150. C14.280. C16.131.
Laryngocele	C16.131.740.650	C8.360. C9.400.	C8.695.
Laryngostenosis	C16.131.740.658	C8.360.	C9.400.
Scimitar Syndrome	C16.131.740.815	C8.381. C14.240. C16.131.	C8.695. C14.907.
Tracheobronchomegaly	C16.131.740.830	C8.127. C8.907.	C8.695.
Situs Inversus	C16.131.810		
Dextrocardia	C16.131.810.250	C14.240. C16.131.	C14.280.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities			
Congenital Abnormalities			
Situs Inversus			
Dextrocardia			
Kartagener Syndrome			
Kartagener Syndrome	C16.131.810.250.500	C8.127. C8.695. C14.240. C16.131. C16.320.	C8.200. C9.150. C14.280. C16.131.
Levocardia	C16.131.810.700	C14.240. C16.131.	C14.280.
Skin Abnormalities	C16.131.831	C17.800.	
Acrodermatitis	C16.131.831.66	C17.800.	C17.800.
Carney Complex	C16.131.831.108	C4.557. C14.280.	C4.588. C16.131.
Dyskeratosis Congenita	C16.131.831.150	C16.320. C17.800.	C16.320. C17.800.
Ectodermal Dysplasia	C16.131.831.350	C16.131. C17.800.	C16.320. C17.800.
Ectodermal Dysplasia 1, Anhidrotic	C16.131.831.350.198	C16.131. C16.320. C17.800.	C16.320. C17.800.
Ectodermal Dysplasia 3, Anhidrotic	C16.131.831.350.298	C16.131. C17.800.	C16.320. C17.800.
Ectodermal Dysplasia, Hypohidrotic, Autosomal Recessive	C16.131.831.350.348	C16.131. C17.800.	C16.320. C17.800.
Ellis-Van Creveld Syndrome	C16.131.831.350.398	C5.116. C16.320. C17.800.	C16.131. C17.800.
Focal Dermal Hypoplasia	C16.131.831.350.424	C5.116. C16.320. C17.800.	C16.131. C16.320. C17.800.
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.
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.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities			
Congenital Abnormalities			
Skin Abnormalities			
Ichthyosis			
Ichthyosiform Erythroderma, Congenital			
Netherton Syndrome			
Netherton Syndrome	C16.131.831.512.400.705	C16.131. C16.320. C17.800. C17.800.	C16.320. C16.614. C17.800. 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. 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.	
Prolidase Deficiency	C16.131.831.720	C16.131. C16.320.	C16.320.
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.
Stomatognathic System Abnormalities	C16.131.850	C7.650	
Maxillofacial Abnormalities	C16.131.850.500	C5.660. C16.131.	C7.650.
Jaw Abnormalities	C16.131.850.500.460	C5.500. C7.320. C16.131.	C5.660. C7.650.
Cleft Palate	C16.131.850.500.460.185	C5.500. C7.320. C7.650. C16.131.	C5.660. C7.465. C7.650. C16.131.
Micrognathism	C16.131.850.500.460.457	C5.500. C7.320. C16.131.	C5.660. C7.650.
Pierre Robin Syndrome	C16.131.850.500.460.606	C5.500. C7.320. C16.131.	C5.660. C7.650.
Prognathism	C16.131.850.500.460.655	C5.500. C5.660. C7.320. C16.131.	C5.500. C7.320. C7.650.
Retrognathism	C16.131.850.500.460.813	C5.500. C5.660. C7.320. C16.131.	C5.500. C7.320. C7.650.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Congenital Abnormalities
Stomatognathic System Abnormalities
Mouth Abnormalities

Mouth Abnormalities	C16.131.850.525	C7.465.	C7.650.
Cleft Lip	C16.131.850.525.164	C7.465.	C7.465.
		C7.650.	
Cleft Palate	C16.131.850.525.185	C5.500.	C5.660.
		C7.320.	C7.465.
		C7.650.	C7.650.
		C16.131.	C16.131.
Fibromatosis, Gingival	C16.131.850.525.304	C7.465.	C7.465.
		C7.650.	
Macrostomia	C16.131.850.525.480	C7.465.	C7.650.
Microstomia	C16.131.850.525.520	C7.465.	C7.650.
Velopharyngeal Insufficiency	C16.131.850.525.955	C7.465.	C7.550.
		C7.650.	C9.775.
Tooth Abnormalities	C16.131.850.800	C7.650.	C7.793.
Anodontia	C16.131.850.800.100	C7.650.	C7.793.
Dens in Dente	C16.131.850.800.250	C7.650.	C7.793.
Dental Enamel Hypoplasia	C16.131.850.800.255	C7.650.	C7.793.
Amelogenesis Imperfecta	C16.131.850.800.255.500	C7.650.	C7.793.
Dentin Dysplasia	C16.131.850.800.260	C7.650.	C7.793.
Dentinogenesis Imperfecta	C16.131.850.800.270	C7.650.	C7.793.
Fused Teeth	C16.131.850.800.370	C7.650.	C7.793.
Odontodysplasia	C16.131.850.800.600	C7.650.	C7.793.
Tooth, Supernumerary	C16.131.850.800.850	C7.650.	C7.793.
Thyroid Dysgenesis	C16.131.894	C19.874.	
Lingual Thyroid	C16.131.894.500	C19.874.	
Lingual Goiter	C16.131.894.500.500	C19.874.	C19.874.
Urogenital Abnormalities	C16.131.939	C12.706	C13.351.
Bladder Exstrophy	C16.131.939.132	C12.706.	C12.777.
		C13.351.	C13.351.
Cryptorchidism	C16.131.939.258	C12.294.	C12.706.
		C19.391.	
Disorders of Sex Development	C16.131.939.316	C12.706.	C13.351.
		C19.391.	F3.800.
46, XX Disorders of Sex Development	C16.131.939.316.64	C12.706.	C13.351.
		C19.391.	F3.800.
46, XX Testicular Disorders of Sex Development	C16.131.939.316.64.124	C12.706.	C13.351.
		C19.391.	
Gonadal Dysgenesis, 46,XX	C16.131.939.316.64.249	C12.706.	C12.706.
		C13.351.	C13.351.
		C16.131.	C19.391.
		C19.391.	
Hyperandrogenism	C16.131.939.316.64.500	C12.706.	C12.706.
		C13.351.	C13.351.
		C16.131.	C19.391.
		C19.391.	
46, XY Disorders of Sex Development	C16.131.939.316.96	C12.706.	C13.351.
		C19.391.	F3.800.
Androgen-Insensitivity Syndrome	C16.131.939.316.96.500	C12.706.	C13.351.
		C16.320.	C19.391.
Denys-Drash Syndrome	C16.131.939.316.96.562	C4.557.	C4.588.
		C4.700.	C12.706.
		C12.758.	C12.777.
		C13.351.	C13.351.
		C13.351.	C16.320.
		C19.391.	
Frasier Syndrome	C16.131.939.316.96.624	C12.706.	C12.777.
		C13.351.	C13.351.
		C16.320.	C19.391.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Congenital Abnormalities
Urogenital Abnormalities
Disorders of Sex Development
46, XY Disorders of Sex Development
Gonadal Dysgenesis, 46,XY

Gonadal Dysgenesis, 46,XY	C16.131.939.316.96.687	C12.706. C13.351. C16.131. C19.391.	C12.706. C13.351. C19.391.
Gonadoblastoma	C16.131.939.316.96.687.500	C4.557. C12.706. C13.351. C16.131. C19.391.	C4.557. C12.706. C13.351. C19.391.
Kallmann Syndrome	C16.131.939.316.96.750	C12.706. C16.320. C19.391.	C13.351. C19.391.
WAGR Syndrome	C16.131.939.316.96.875	C4.557. C4.700. C11.250. C11.941. C12.758. C13.351. C13.351. C16.131. C16.320. C19.391.	C4.588. C10.597. C11.270. C12.706. C12.777. C13.351. C16.131. C16.320. C16.320.
Adrenogenital Syndrome	C16.131.939.316.129	C12.706. C19.391.	C13.351.
Adrenal Hyperplasia, Congenital	C16.131.939.316.129.500	C12.706. C16.320. C18.452. C19.391.	C13.351. C16.320. C19.53.
Hyperandrogenism	C16.131.939.316.129.750	C12.706. C13.351. C16.131. C19.391.	C12.706. C13.351. C19.391.
Gonadal Dysgenesis	C16.131.939.316.309	C12.706. C19.391.	C13.351.
Gonadal Dysgenesis, 46,XX	C16.131.939.316.309.193	C12.706. C13.351. C16.131. C19.391.	C12.706. C13.351. C19.391.
Gonadal Dysgenesis, 46,XY	C16.131.939.316.309.388	C12.706. C13.351. C16.131. C19.391.	C12.706. C13.351. C19.391.
Gonadoblastoma	C16.131.939.316.309.388.500	C4.557. C12.706. C13.351. C16.131. C19.391.	C4.557. C12.706. C13.351. C19.391.
Gonadal Dysgenesis, Mixed	C16.131.939.316.309.391	C12.706. C13.351. C16.131. C16.320. C19.391.	C12.706. C13.351. C16.131. C19.391.
Sexual Infantilism	C16.131.939.316.309.631	C12.706. C19.391.	C13.351. C19.391.
Turner Syndrome	C16.131.939.316.309.872	C12.706. C13.351. C14.240. C16.131. C16.131. C19.391.	C12.706. C13.351. C14.280. C16.131. C16.320. C19.391.

C16 - DISEASES-CONGENITAL AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities			
Congenital Abnormalities			
Urogenital Abnormalities			
Disorders of Sex Development			
Ovotesticular Disorders of Sex Development			
Ovotesticular Disorders of Sex Development	C16.131.939.316.343	C12.706.	C13.351.
		C19.391.	
Sex Chromosome Disorders of Sex Development	C16.131.939.316.795	C12.706.	C13.351.
		C16.131.	C16.320.
		C19.391.	
Freemartinism	C16.131.939.316.795.124	C12.706.	C13.351.
		C19.391.	C22.196.
Gonadal Dysgenesis, Mixed	C16.131.939.316.795.249	C12.706.	C12.706.
		C13.351.	C13.351.
		C16.131.	C16.131.
		C16.320.	C19.391.
		C19.391.	
Klinefelter Syndrome	C16.131.939.316.795.500	C12.706.	C13.351.
		C16.131.	C16.320.
		C19.391.	C19.391.
Turner Syndrome	C16.131.939.316.795.750	C12.706.	C12.706.
		C13.351.	C13.351.
		C14.240.	C14.280.
		C16.131.	C16.131.
		C16.131.	C16.320.
		C19.391.	C19.391.
Epispadias	C16.131.939.374	C12.706.	C12.777.
		C13.351.	C13.351.
Hypospadias	C16.131.939.516	C12.294.	C12.706.
		C13.351.	
Multicystic Dysplastic Kidney	C16.131.939.629	C12.706.	C12.777.
		C13.351.	C13.351.
Nephritis, Hereditary	C16.131.939.742	C12.706.	C12.777.
		C13.351.	C13.351.
		C17.300.	
Pyelectasis	C16.131.939.831	C13.703.	C16.300.
Fetal Diseases	C16.300	C13.703.	
Chorioamnionitis	C16.300.30	C13.703.	C13.703.
		C13.703.	
Echogenic Bowel	C16.300.50	C13.703.	
Erythroblastosis, Fetal	C16.300.60	C13.703.	C15.378.
		C16.614.	C20.188.
Hydrops Fetalis	C16.300.60.480	C13.703.	C15.378.
		C15.378.	C16.320.
		C20.188.	C23.888.
Fetal Alcohol Syndrome	C16.300.80	C13.703.	C25.100.
Fetal Growth Retardation	C16.300.390	C13.703.	C23.550.
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.	
Pyelectasis	C16.300.895	C13.703.	C16.131.
Genetic Diseases, Inborn	C16.320		
Adrenal Hyperplasia, Congenital	C16.320.33	C12.706.	C13.351.
		C16.131.	C16.320.
		C18.452.	C19.53.
		C19.391.	
Alagille Syndrome	C16.320.51	C6.130.	C6.552.
		C14.240.	C16.131.
		C16.131.	
alpha 1-Antitrypsin Deficiency	C16.320.60	C6.552.	C8.381.
		C23.550.	

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Anemia, Hemolytic, Congenital

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.
Acute Chest Syndrome	C16.320.70.150.219	C16.320. C8.381. C15.378.	C8.618. C15.378.
Hemoglobin SC Disease	C16.320.70.150.440	C16.320. C15.378.	C15.378.
Sickle Cell Trait	C16.320.70.150.670	C16.320. C15.378.	C15.378.
Elliptocytosis, Hereditary	C16.320.70.365	C16.320. C15.378.	
Glucosephosphate Dehydrogenase Deficiency	C16.320.70.480	C15.378.	C16.320.
Favism	C16.320.70.480.370	C18.452. C15.378. C25.723.	C15.378. C25.723.
Hemoglobin C Disease	C16.320.70.490	C15.378.	C15.378.
Spherocytosis, Hereditary	C16.320.70.785	C16.320. C15.378.	
Thalassemia	C16.320.70.875	C15.378.	C15.378.
alpha-Thalassemia	C16.320.70.875.100	C16.320. C15.378.	C15.378.
beta-Thalassemia	C16.320.70.875.150	C16.320. C15.378.	C15.378.
delta-Thalassemia	C16.320.70.875.575	C16.320. C15.378.	C15.378.
Anemia, Hypoplastic, Congenital	C16.320.77	C16.320. C15.378.	C15.378.
Anemia, Diamond-Blackfan	C16.320.77.90	C15.378.	C15.378.
Fanconi Anemia	C16.320.77.280	C15.378. C15.378.	C15.378.
Angioedemas, Hereditary	C16.320.78	C18.452. C14.907.	C17.800.
Ataxia Telangiectasia	C16.320.80	C20.543. C10.228. C10.597. C18.452.	C10.562. C14.907. C20.673.
Autoimmune Lymphoproliferative Syndrome	C16.320.89	C15.604. C20.683.	C20.111.
Blood Coagulation Disorders, Inherited	C16.320.99	C15.378.	
Activated Protein C Resistance	C16.320.99.37	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.
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.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities			
Genetic Diseases, Inborn			
Blood Coagulation Disorders, Inherited			
Factor XIII Deficiency			
Factor XIII Deficiency	C16.320.99.335	C15.378.	C15.378.
		C15.378.	
Gray Platelet Syndrome	C16.320.99.417	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.	C16.320.
Hermanski-Pudlak Syndrome	C16.320.99.515	C11.270.	C15.378.
		C15.378.	C15.378.
		C15.378.	C16.320.
		C16.320.	C16.320.
		C17.800.	C17.800.
		C18.452.	
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 Diseases	C16.320.99.920	C15.378.	C15.378.
		C15.378.	C15.378.
von Willebrand Disease, Type 1	C16.320.99.920.100	C15.378.	C15.378.
		C15.378.	
von Willebrand Disease, Type 2	C16.320.99.920.200	C15.378.	C15.378.
		C15.378.	
von Willebrand Disease, Type 3	C16.320.99.920.300	C15.378.	C15.378.
		C15.378.	
Wiskott-Aldrich Syndrome	C16.320.99.970	C15.378.	C15.378.
		C15.378.	C16.320.
		C20.673.	C20.673.
Brugada Syndrome	C16.320.100	C14.280.	
CADASIL	C16.320.129	C10.228.	C10.228.
		C10.228.	C10.228.
		C14.907.	C14.907.
Camurati-Engelmann Syndrome	C16.320.144	C5.116.	
Cardiomyopathy, Hypertrophic, Familial	C16.320.160	C14.280.	
CHARGE Syndrome	C16.320.165	C16.131.	
Cherubism	C16.320.170	C5.116.	C5.500.
		C7.320.	C16.131.
Chromosome Disorders	C16.320.180	C16.131.	
22q11 Deletion Syndrome	C16.320.180.19	C5.660.	C14.240.
		C14.280.	C15.604.
		C16.131.	C16.131.
		C16.131.	C16.131.
		C16.131.	C19.642.
DiGeorge Syndrome	C16.320.180.19.500	C5.660.	C14.240.
		C14.280.	C15.604.
		C16.131.	C16.131.
		C16.131.	C16.131.
		C16.131.	C19.642.
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.	

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Chromosome Disorders
Holoprosencephaly

Holoprosencephaly	C16.320.180.380	C5.660.	C10.500.
		C16.131.	C16.131.
		C16.131.	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.	C5.660.
		C10.597.	C16.131.
		C16.131.	C16.131.
Sex Chromosome Disorders	C16.320.180.830	C16.131.	
Fragile X Syndrome	C16.320.180.830.300	C10.597.	C16.131.
		C16.320.	C16.320.
Orofaciodigital Syndromes	C16.320.180.830.670	C5.116.	C5.660.
		C16.131.	C16.131.
		C16.131.	
Sex Chromosome Disorders of Sex Development	C16.320.180.830.835	C12.706.	C13.351.
		C16.131.	C16.131.
		C19.391.	
Gonadal Dysgenesis, Mixed	C16.320.180.830.835.249	C12.706.	C12.706.
		C13.351.	C13.351.
		C16.131.	C16.131.
		C16.131.	C19.391.
		C19.391.	
Klinefelter Syndrome	C16.320.180.830.835.500	C12.706.	C13.351.
		C16.131.	C16.131.
		C19.391.	C19.391.
Turner Syndrome	C16.320.180.830.835.750	C12.706.	C12.706.
		C13.351.	C13.351.
		C14.240.	C14.280.
		C16.131.	C16.131.
		C16.131.	C16.131.
		C19.391.	C19.391.
Silver-Russell Syndrome	C16.320.180.870	C5.660.	C16.131.
		C16.131.	C16.320.
Smith-Magenis Syndrome	C16.320.180.887	C10.281.	C16.131.
		C16.131.	
Sotos Syndrome	C16.320.180.905	C16.131.	C16.131.
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.
		C19.391.	
Williams Syndrome	C16.320.180.970	C10.597.	C14.280.
		C16.131.	
Wolf-Hirschhorn Syndrome	C16.320.180.985	C16.131.	C16.131.
Costello Syndrome	C16.320.185	C5.660.	C16.131.
Cystic Fibrosis	C16.320.190	C6.689.	C8.381.
		C16.614.	
Donohue Syndrome	C16.320.215	C5.660.	C16.131.
		C18.452.	C19.246.
Dwarfism	C16.320.240	C5.116.	C19.297
Achondroplasia	C16.320.240.500	C5.116.	C5.116.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities			
Genetic Diseases, Inborn			
Dwarfism			
Achondroplasia			
Thanatophoric Dysplasia			
Thanatophoric Dysplasia	C16.320.240.500.500	C5.116. C5.660. C16.614.	C5.116. C16.131.
Cockayne Syndrome	C16.320.240.562	C5.116. C16.131. C18.452.	C10.574. C16.320.
Congenital Hypothyroidism	C16.320.240.625	C5.116. C19.297.	C5.116. C19.874.
Laron Syndrome	C16.320.240.750	C5.116.	C19.297.
Mulibrey Nanism	C16.320.240.875	C5.116.	
Silver-Russell Syndrome	C16.320.240.937	C5.660. C16.131.	C16.131. C16.320.
Eye Diseases, Hereditary	C16.320.290	C11.270	
Aicardi Syndrome	C16.320.290.19	C10.500. C16.131. C16.320.	C11.270. C16.131.
Albinism	C16.320.290.40	C11.270. C16.320. C17.800.	C16.320. C17.800. C18.452.
Albinism, Ocular	C16.320.290.40.90	C11.270. C16.320. C17.800.	C16.320. C17.800. C18.452.
Albinism, Oculocutaneous	C16.320.290.40.100	C11.270. C16.320. C17.800.	C16.320. C17.800. C18.452.
Hermanski-Pudlak Syndrome	C16.320.290.40.100.400	C11.270. C15.378. C15.378. C16.320. C17.800. C18.452.	C15.378. C15.378. C16.320. C16.320. C17.800.
Piebaldism	C16.320.290.40.600	C16.320. C17.800. C18.452.	C16.320. C17.800.
Aniridia	C16.320.290.78	C11.250. C11.941.	C11.270. C16.131.
WAGR Syndrome	C16.320.290.78.950	C4.557. C4.700. C11.250. C11.941. C12.758. C13.351. C13.351. C16.131. C16.320. C19.391.	C4.588. C10.597. C11.270. C12.706. C12.777. C13.351. C16.131. C16.131. C16.320.
Choroideremia	C16.320.290.142	C11.270. C16.320.	C11.941.
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. C11.590.	C11.270.
Gyrate Atrophy	C16.320.290.468	C11.270.	C11.941.
Optic Atrophies, Hereditary	C16.320.290.564	C10.292. C11.270. C16.320.	C10.574. C11.640.
Optic Atrophy, Autosomal Dominant	C16.320.290.564.100	C10.292. C11.270. C16.320.	C10.574. C11.640. C18.452.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Eye Diseases, Hereditary
Optic Atrophies, Hereditary
Optic Atrophy, Hereditary, Leber

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.
Retinal Dysplasia	C16.320.290.660	C11.250.	C11.270.
		C11.768.	C16.131.
Retinitis Pigmentosa	C16.320.290.684	C11.270.	C11.768.
Alstrom Syndrome	C16.320.290.684.249	C10.500.	C10.574.
		C10.668.	C11.270.
		C16.131.	C16.131.
		C16.320.	
Usher Syndromes	C16.320.290.684.500	C9.218.	C9.218.
		C10.597.	C10.597.
		C10.597.	C11.768.
		C11.966.	C16.131.
		C23.888.	
Vitelliform Macular Dystrophy	C16.320.290.763	C11.768.	
Weill-Marchesani Syndrome	C16.320.290.842	C5.116.	C5.116.
		C11.270.	C16.131.
		C16.320.	C17.300.
Frasier Syndrome	C16.320.306	C12.706.	C12.777.
		C13.351.	C13.351.
		C16.131.	C19.391.
Genetic Diseases, X-Linked	C16.320.322		
Aicardi Syndrome	C16.320.322.30	C10.500.	C11.270.
		C16.131.	C16.131.
		C16.320.	
Androgen-Insensitivity Syndrome	C16.320.322.61	C12.706.	C13.351.
		C16.131.	C19.391.
Barth Syndrome	C16.320.322.68	C14.240.	C14.280.
		C16.131.	C16.131.
		C16.320.	C18.452.
Bulbo-Spinal Atrophy, X-Linked	C16.320.322.76	C10.228.	C10.574.
		C10.574.	C10.668.
Choroideremia	C16.320.322.92	C11.270.	C11.941.
		C16.320.	
Dent Disease	C16.320.322.100	C12.777.	C13.351.
		C16.320.	C18.452.
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.	C10.228.
		C14.907.	C16.320.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Focal Dermal Hypoplasia	C16.320.322.186	C5.116.	C16.131.
		C16.131.	C16.320.
		C17.800.	C17.800.
Glycogen Storage Disease Type IIb	C16.320.322.201	C10.597.	C14.280.
		C16.320.	C18.452.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Genetic Diseases, X-Linked
Glycogen Storage Disease Type VIII

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.	C12.777.
		C13.351.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.654.	
Ichthyosis, X-Linked	C16.320.322.241	C16.131.	C16.320.
		C16.320.	C16.614.
		C17.800.	C17.800.
		C17.800.	C18.452.
Isolated Noncompaction of the Ventricular Myocardium	C16.320.322.370	C14.240.	C14.280.
		C14.280.	C16.131.
		C16.131.	
Mental Retardation, X-Linked	C16.320.322.500	C10.597.	C16.320.
Adrenoleukodystrophy	C16.320.322.500.124	C10.228.	C10.228.
		C10.228.	C10.228.
		C10.314.	C10.597.
		C16.320.	C16.320.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	C19.53.
Classical Lissencephalies and Subcortical Band Heterotopias	C16.320.322.500.186	C10.500.	C10.500.
		C16.131.	C16.131.
Coffin-Lowry Syndrome	C16.320.322.500.249	C10.597.	C16.320.
Fragile X Syndrome	C16.320.322.500.500	C10.597.	C16.131.
		C16.320.	C16.320.
Lesch-Nyhan Syndrome	C16.320.322.500.625	C10.228.	C10.574.
		C10.597.	C16.320.
		C16.320.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
Menkes Kinky Hair Syndrome	C16.320.322.500.687	C10.228.	C10.574.
		C10.597.	C16.320.
		C16.320.	C16.320.
		C16.320.	C17.800.
		C18.452.	C18.452.
		C18.452.	
Mucopolysaccharidosis II	C16.320.322.500.750	C10.597.	C16.320.
		C16.320.	C16.320.
		C17.300.	C18.452.
		C18.452.	
Pyruvate Dehydrogenase Complex Deficiency Disease	C16.320.322.500.875	C10.228.	C10.597.
		C16.320.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Rett Syndrome	C16.320.322.500.937	C10.574.	C10.597.
		C16.320.	C16.320.
Muscular Dystrophy, Duchenne	C16.320.322.562	C5.651.	C10.668.
		C16.320.	
Muscular Dystrophy, Emery-Dreifuss	C16.320.322.625	C5.651.	C10.668.
		C16.320.	

Congenital, Hereditary, and Neonatal Diseases and Abnormalities			
Genetic Diseases, Inborn			
Genetic Diseases, X-Linked			
Oculocerebrorenal Syndrome			
Oculocerebrorenal Syndrome	C16.320.322.750	C10.228.	C12.777.
		C13.351.	C16.131.
		C16.320.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Ornithine Carbamoyltransferase Deficiency Disease	C16.320.322.828	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
Pelizaeus-Merzbacher Disease	C16.320.322.906	C10.228.	C10.228.
		C10.228.	C10.314.
		C16.320.	C18.452.
		C18.452.	
Wiskott-Aldrich Syndrome	C16.320.322.937	C15.378.	C15.378.
		C15.378.	C16.320.
		C20.673.	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.	
Acute Chest Syndrome	C16.320.365.155.219	C8.381.	C8.618.
		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.	
delta-Thalassemia	C16.320.365.826.575	C15.378.	C15.378.
		C16.320.	
Hereditary Autoinflammatory Diseases	C16.320.382	C17.800.	
Cryopyrin-Associated Periodic Syndromes	C16.320.382.500	C17.800.	
Familial Mediterranean Fever	C16.320.382.625		
Mevalonate Kinase Deficiency	C16.320.382.750	C10.228.	C15.378.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C20.683.
Heredodegenerative Disorders, Nervous System	C16.320.400	C10.574.	
Alexander Disease	C16.320.400.24	C10.228.	C10.228.
		C10.228.	C10.314.
		C10.574.	C16.320.
		C18.452.	C18.452.
Amyloid Neuropathies, Familial	C16.320.400.50	C10.574.	C10.668.
		C16.320.	C18.452.
		C18.452.	C18.452.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Heredodegenerative Disorders, Nervous System
Canavan Disease

Canavan Disease	C16.320.400.150	C10.228. C10.228. C10.574. C18.452.	C10.228. C10.314. C16.320. C18.452.
Cockayne Syndrome	C16.320.400.200	C5.116. C16.131. C18.452.	C10.574. C16.320.
Dystonia Musculorum Deformans	C16.320.400.330	C10.228. C10.574.	C10.228.
Gerstmann-Straussler-Scheinker Disease	C16.320.400.350	C10.228. C10.574.	C10.574.
Hepatolenticular Degeneration	C16.320.400.361	C6.552. C10.228. C10.574. C16.320. C18.452.	C10.228. C10.228. C16.320. C18.452. C18.452.
Hereditary Central Nervous System Demyelinating Diseases	C16.320.400.367	C10.228. C10.228. C10.574. C18.452.	C10.228. C10.314. C16.320. C18.452.
Hereditary Sensory and Motor Neuropathy	C16.320.400.375	C10.500. C10.668.	C10.574. C16.131.
Alstrom Syndrome	C16.320.400.375.99	C10.500. C10.668. C16.131. C16.320.	C10.574. C11.270. C16.131.
Charcot-Marie-Tooth Disease	C16.320.400.375.200	C10.500. C10.668.	C10.574. C16.131.
Giant Axonal Neuropathy	C16.320.400.375.490	C10.500. C10.668. C16.131.	C10.574. C10.668.
Refsum Disease	C16.320.400.375.780	C10.228. C10.574. C16.131. C16.320. C18.452.	C10.500. C10.668. C16.320. C18.452. C18.452.
Spastic Paraplegia, Hereditary	C16.320.400.375.820	C10.500. C10.668.	C10.574. C16.131.
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. C18.452.	C10.574. C16.320. C16.320. C18.452. C18.452.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Heredodegenerative Disorders, Nervous System
Menkes Kinky Hair Syndrome

Menkes Kinky Hair Syndrome	C16.320.400.520	C10.228.	C10.574.
		C10.597.	C16.320.
		C16.320.	C16.320.
		C16.320.	C17.800.
		C18.452.	C18.452.
		C18.452.	
Mental Retardation, X-Linked	C16.320.400.525	C10.597.	C16.320.
Adrenoleukodystrophy	C16.320.400.525.124	C10.228.	C10.228.
		C10.228.	C10.228.
		C10.314.	C10.597.
		C16.320.	C16.320.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	C19.53.
Coffin-Lowry Syndrome	C16.320.400.525.249	C10.597.	C16.320.
Fragile X Syndrome	C16.320.400.525.500	C10.597.	C16.131.
		C16.320.	C16.320.
Lesch-Nyhan Syndrome	C16.320.400.525.625	C10.228.	C10.574.
		C10.597.	C16.320.
		C16.320.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
Menkes Kinky Hair Syndrome	C16.320.400.525.687	C10.228.	C10.574.
		C10.597.	C16.320.
		C16.320.	C16.320.
		C16.320.	C17.800.
		C18.452.	C18.452.
		C18.452.	
Mucopolysaccharidosis II	C16.320.400.525.750	C10.597.	C16.320.
		C16.320.	C16.320.
		C17.300.	C18.452.
		C18.452.	
Pyruvate Dehydrogenase Complex Deficiency Disease	C16.320.400.525.875	C10.228.	C10.597.
		C16.320.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Rett Syndrome	C16.320.400.525.937	C10.574.	C10.597.
		C16.320.	C16.320.
Myotonia Congenita	C16.320.400.540	C5.651.	C10.574.
		C10.668.	
Myotonic Dystrophy	C16.320.400.542	C5.651.	C5.651.
		C10.574.	C10.668.
		C10.668.	C16.320.
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.	C4.700.
		C9.218.	C9.647.
		C10.292.	C10.292.
		C10.562.	C10.574.
		C16.320.	
Neuronal Ceroid-Lipofuscinoses	C16.320.400.600	C10.574.	C16.320.
		C18.452.	C18.452.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Heredodegenerative Disorders, Nervous System
Optic Atrophies, Hereditary

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.
Pantothenate Kinase-Associated Neurodegeneration	C16.320.400.650	C10.228.	C10.228.
		C10.228.	C10.574.
Rett Syndrome	C16.320.400.700	C10.574.	C10.597.
		C16.320.	C16.320.
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.	C4.651.
		C4.700.	C10.500.
		C10.562.	C10.574.
		C16.131.	C16.320.
Unverricht-Lundborg Syndrome	C16.320.400.940	C10.228.	C10.574.
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.	
Loeys-Dietz Syndrome	C16.320.510	C5.660.	C14.907.
		C14.907.	C14.907.
		C16.131.	

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Marfan Syndrome

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.	
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.	C18.452.
		C18.654.	
Homocystinuria	C16.320.565.100.480.500	C10.228.	C16.320.
		C17.300.	C18.452.
		C18.452.	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.	
Holocarboxylase Synthetase Deficiency	C16.320.565.100.620.380	C16.320.	C18.452.
		C18.452.	
Phenylketonurias	C16.320.565.100.766	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Phenylketonuria, Maternal	C16.320.565.100.766.500	C10.228.	C13.703.
		C16.320.	C18.452.
		C18.452.	C18.452.
Prolidase Deficiency	C16.320.565.100.794	C16.131.	C16.131.
		C16.320.	
Propionic Acidemia	C16.320.565.100.823	C18.452.	
Tyrosinemias	C16.320.565.100.880	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Urea Cycle Disorders, Inborn	C16.320.565.100.940	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	

Congenital, Hereditary, and Neonatal Diseases and Abnormalities			
Genetic Diseases, Inborn			
Metabolism, Inborn Errors			
Amino Acid Metabolism, Inborn Errors			
Urea Cycle Disorders, Inborn			
Argininosuccinic Aciduria			
Argininosuccinic Aciduria	C16.320.565.100.940.124	C10.228. C18.452. C18.452.	C16.320. C18.452.
Carbamoyl-Phosphate Synthase I Deficiency Disease	C16.320.565.100.940.249	C10.228. C18.452. C18.452.	C16.320. C18.452. C18.452.
Citrullinemia	C16.320.565.100.940.374	C10.228. C18.452. C18.452.	C16.320. C18.452.
Hyperargininemia	C16.320.565.100.940.500	C10.228. C18.452. C18.452.	C16.320. C18.452.
Ornithine Carbamoyltransferase Deficiency Disease	C16.320.565.100.940.750	C10.228. C16.320. C18.452.	C16.320. C18.452. C18.452.
Amino Acid Transport Disorders, Inborn	C16.320.565.151	C18.452.	
Hartnup Disease	C16.320.565.151.355	C10.228. C13.351. C16.320. C18.452.	C12.777. C16.320. C18.452. C18.452.
Oculocerebrorenal Syndrome	C16.320.565.151.600	C10.228. C13.351. C16.320. C16.320. C18.452. C18.452.	C12.777. C16.131. C16.320. C18.452. C18.452.
Amyloidosis, Familial	C16.320.565.176	C18.452.	C18.452.
Amyloid Neuropathies, Familial	C16.320.565.176.50	C10.574. C16.320. C18.452.	C10.668. C18.452. C18.452.
Cerebral Amyloid Angiopathy, Familial	C16.320.565.176.160	C10.228. C10.228. C14.907. C18.452. C18.452. C18.452.	C10.228. C14.907. C16.320. C18.452. C18.452.
Brain Diseases, Metabolic, Inborn	C16.320.565.189	C10.228. C18.452.	C18.452.
Cerebral Amyloid Angiopathy, Familial	C16.320.565.189.168	C10.228. C10.228. C14.907. C18.452. C18.452. C18.452.	C10.228. C14.907. C16.320. C18.452. C18.452.
Galactosemias	C16.320.565.189.320	C10.228. C18.452. C18.452.	C16.320. C18.452.
Hartnup Disease	C16.320.565.189.355	C10.228. C13.351. C16.320. C18.452.	C12.777. C16.320. C18.452. C18.452.
Hepatolenticular Degeneration	C16.320.565.189.360	C6.552. C10.228. C10.574. C16.320. C18.452.	C10.228. C10.228. C16.320. C18.452. C18.452.
Hereditary Central Nervous System Demyelinating Diseases	C16.320.565.189.362	C10.228. C10.228. C10.574. C18.452.	C10.228. C10.314. C16.320. C18.452.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Metabolism, Inborn Errors
Brain Diseases, Metabolic, Inborn
Hereditary Central Nervous System Demyelinating Diseases
Adrenoleukodystrophy

Adrenoleukodystrophy	C16.320.565.189.362.250	C10.228.	C10.228.
		C10.228.	C10.228.
		C10.314.	C10.597.
		C16.320.	C16.320.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	C19.53.
Alexander Disease	C16.320.565.189.362.312	C10.228.	C10.228.
		C10.228.	C10.314.
		C10.574.	C16.320.
		C18.452.	C18.452.
Canavan Disease	C16.320.565.189.362.375	C10.228.	C10.228.
		C10.228.	C10.314.
		C10.574.	C16.320.
		C18.452.	C18.452.
Leukodystrophy, Globoid Cell	C16.320.565.189.362.500	C10.228.	C10.228.
		C10.228.	C10.228.
		C10.314.	C16.320.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Leukodystrophy, Metachromatic	C16.320.565.189.362.550	C10.228.	C10.228.
		C10.228.	C10.228.
		C10.314.	C16.320.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Pelizaeus-Merzbacher Disease	C16.320.565.189.362.775	C10.228.	C10.228.
		C10.228.	C10.314.
		C16.320.	C18.452.
		C18.452.	
Homocystinuria	C16.320.565.189.365	C10.228.	C16.320.
		C17.300.	C18.452.
		C18.452.	C18.452.
Hyperglycinemia, Nonketotic	C16.320.565.189.375	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Hyperlysinemias	C16.320.565.189.380	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Leigh Disease	C16.320.565.189.412	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
Lesch-Nyhan Syndrome	C16.320.565.189.425	C10.228.	C10.574.
		C10.597.	C16.320.
		C16.320.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
Lysosomal Storage Diseases, Nervous System	C16.320.565.189.435	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Fucosidosis	C16.320.565.189.435.295	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Metabolism, Inborn Errors
Brain Diseases, Metabolic, Inborn
Lysosomal Storage Diseases, Nervous System
Glycogen Storage Disease Type II

Glycogen Storage Disease Type II	C16.320.565.189.435.340	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452. C18.452.
Mucolipidoses	C16.320.565.189.435.590	C5.116. C16.320. C18.452. C18.452.	C10.228. C16.320. C18.452. C18.452.
Sialic Acid Storage Disease	C16.320.565.189.435.810	C10.228. C18.452. C18.452.	C16.320. C18.452.
Sphingolipidoses	C16.320.565.189.435.825	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452. C18.452. C18.452.
Fabry Disease	C16.320.565.189.435.825.200	C10.228. C14.907. C16.320. C18.452. C18.452. C18.452.	C10.228. C16.320. C16.320. C18.452. C18.452.
Farber Lipogranulomatosis	C16.320.565.189.435.825.250	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452. C18.452. C18.452.
Gangliosidoses	C16.320.565.189.435.825.300	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452. C18.452. C18.452.
Gangliosidoses, GM2	C16.320.565.189.435.825.300.300	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452. C18.452. C18.452.
Sandhoff Disease	C16.320.565.189.435.825.300.300.249	C10.228. C16.320. C16.320. C18.452. C18.452. C18.452.	C10.228. C16.320. C18.452. C18.452. C18.452. C18.452.
Tay-Sachs Disease	C16.320.565.189.435.825.300.300.500	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452. 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.	C16.320. C18.452. C18.452. C18.452.
Gangliosidosis, GM1	C16.320.565.189.435.825.300.400	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452. 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. C18.452. C18.452.
Gaucher Disease	C16.320.565.189.435.825.400	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452. C18.452. C18.452.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Metabolism, Inborn Errors
Brain Diseases, Metabolic, Inborn
Lysosomal Storage Diseases, Nervous System
Sphingolipidoses

Leukodystrophy, Globoid Cell	C16.320.565.189.435.825.590	C10.228.	C10.228.
		C10.228.	C10.228.
		C10.314.	C16.320.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Niemann-Pick Diseases	C16.320.565.189.435.825.700	C10.228.	C15.604.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Niemann-Pick Disease, Type A	C16.320.565.189.435.825.700.500	C10.228.	C15.604.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Niemann-Pick Disease, Type B	C16.320.565.189.435.825.700.750	C10.228.	C15.604.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Niemann-Pick Disease, Type C	C16.320.565.189.435.825.700.875	C10.228.	C15.604.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Sea-Blue Histiocyte Syndrome	C16.320.565.189.435.825.775	C10.228.	C15.604.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Sulfatidosis	C16.320.565.189.435.825.850	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
Leukodystrophy, Metachromatic	C16.320.565.189.435.825.850.500	C10.228.	C10.228.
		C10.228.	C10.228.
		C10.314.	C16.320.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Multiple Sulfatase Deficiency Disease	C16.320.565.189.435.825.850.750	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
Maple Syrup Urine Disease	C16.320.565.189.520	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	
MELAS Syndrome	C16.320.565.189.535	C5.651.	C10.228.
		C10.228.	C10.668.
		C14.907.	C18.452.
		C18.452.	C18.452.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Metabolism, Inborn Errors
Brain Diseases, Metabolic, Inborn
Menkes Kinky Hair Syndrome

Menkes Kinky Hair Syndrome	C16.320.565.189.540	C10.228.	C10.574.
		C10.597.	C16.320.
		C16.320.	C16.320.
		C16.320.	C17.800.
		C18.452.	C18.452.
		C18.452.	
MERRF Syndrome	C16.320.565.189.545	C5.651.	C10.228.
		C10.228.	C10.668.
		C18.452.	C18.452.
		C18.452.	
Oculocerebrorenal Syndrome	C16.320.565.189.640	C10.228.	C12.777.
		C13.351.	C16.131.
		C16.320.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Peroxisomal Disorders	C16.320.565.189.680	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Adrenoleukodystrophy	C16.320.565.189.680.100	C10.228.	C10.228.
		C10.228.	C10.228.
		C10.314.	C10.597.
		C16.320.	C16.320.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	C19.53.
Mevalonate Kinase Deficiency	C16.320.565.189.680.430	C10.228.	C15.378.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C20.683.
Refsum Disease	C16.320.565.189.680.760	C10.228.	C10.500.
		C10.574.	C10.668.
		C16.131.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
Refsum Disease, Infantile	C16.320.565.189.680.865	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Zellweger Syndrome	C16.320.565.189.680.970	C6.552.	C10.228.
		C12.777.	C13.351.
		C16.131.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Phenylketonurias	C16.320.565.189.687	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Phenylketonuria, Maternal	C16.320.565.189.687.500	C10.228.	C13.703.
		C16.320.	C18.452.
		C18.452.	C18.452.
Pyruvate Carboxylase Deficiency Disease	C16.320.565.189.725	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
Pyruvate Dehydrogenase Complex Deficiency Disease	C16.320.565.189.750	C10.228.	C10.597.
		C16.320.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Tyrosinemias	C16.320.565.189.875	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Metabolism, Inborn Errors
Brain Diseases, Metabolic, Inborn
Urea Cycle Disorders, Inborn

Urea Cycle Disorders, Inborn	C16.320.565.189.937	C10.228. C18.452. C18.452.	C16.320. C18.452.
Argininosuccinic Aciduria	C16.320.565.189.937.124	C10.228. C18.452. C18.452.	C16.320. C18.452.
Carbamoyl-Phosphate Synthase I Deficiency Disease	C16.320.565.189.937.249	C10.228. C18.452. C18.452.	C16.320. C18.452. C18.452.
Citrullinemia	C16.320.565.189.937.374	C10.228. C18.452. C18.452.	C16.320. C18.452.
Hyperargininemia	C16.320.565.189.937.500	C10.228. C18.452. C18.452.	C16.320. C18.452.
Ornithine Carbamoyltransferase Deficiency Disease	C16.320.565.189.937.750	C10.228. C16.320. C18.452.	C16.320. C18.452. C18.452.
Carbohydrate Metabolism, Inborn Errors	C16.320.565.202	C18.452.	
Congenital Disorders of Glycosylation	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. 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.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Metabolism, Inborn Errors
Carbohydrate Metabolism, Inborn Errors
Mucolipidoses

Mucolipidoses	C16.320.565.202.670	C5.116.	C10.228.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
Mucopolysaccharidoses	C16.320.565.202.715	C16.320.	C17.300.
		C18.452.	C18.452.
Mucopolysaccharidosis I	C16.320.565.202.715.640	C16.320.	C17.300.
		C18.452.	C18.452.
Mucopolysaccharidosis II	C16.320.565.202.715.645	C10.597.	C16.320.
		C16.320.	C16.320.
		C17.300.	C18.452.
		C18.452.	
Mucopolysaccharidosis III	C16.320.565.202.715.650	C16.320.	C17.300.
		C18.452.	C18.452.
Mucopolysaccharidosis IV	C16.320.565.202.715.655	C16.320.	C17.300.
		C18.452.	C18.452.
Mucopolysaccharidosis VI	C16.320.565.202.715.670	C16.320.	C17.300.
		C18.452.	C18.452.
Mucopolysaccharidosis VII	C16.320.565.202.715.675	C16.320.	C17.300.
		C18.452.	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.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
Pyruvate Carboxylase Deficiency Disease	C16.320.565.202.810.666	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
Pyruvate Dehydrogenase Complex Deficiency Disease	C16.320.565.202.810.766	C10.228.	C10.597.
		C16.320.	C16.320.
		C16.320.	C18.452.
		C18.452.	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.
Barth Syndrome	C16.320.565.398.224	C14.240.	C14.280.
		C16.131.	C16.131.
		C16.320.	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.
		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.	

Congenital, Hereditary, and Neonatal Diseases and Abnormalities			
Genetic Diseases, Inborn			
Metabolism, Inborn Errors			
Lipid Metabolism, Inborn Errors			
Hypolipoproteinemias			
Hypobetalipoproteinemias			
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.	C16.614.
		C18.452.	C18.452.
		C18.452.	
Neuronal Ceroid-Lipofuscinoses	C16.320.565.398.641.509	C10.574.	C16.320.
		C18.452.	C18.452.
Sjogren-Larsson Syndrome	C16.320.565.398.641.723	C16.131.	C16.320.
		C16.614.	C17.800.
		C17.800.	C17.800.
		C18.452.	C18.452.
Sphingolipidoses	C16.320.565.398.641.803	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
Fabry Disease	C16.320.565.398.641.803.300	C10.228.	C10.228.
		C14.907.	C16.320.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Farber Lipogranulomatosis	C16.320.565.398.641.803.325	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
Gangliosidoses	C16.320.565.398.641.803.350	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
Gangliosidoses, GM2	C16.320.565.398.641.803.350.300	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
Sandhoff Disease	C16.320.565.398.641.803.350.300.700	C10.228.	C10.228.
		C16.320.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
Tay-Sachs Disease	C16.320.565.398.641.803.350.300.850	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
Tay-Sachs Disease, AB Variant	C16.320.565.398.641.803.350.300.925	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
Gangliosidosis, GM1	C16.320.565.398.641.803.350.360	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
Gaucher Disease	C16.320.565.398.641.803.441	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities			
Genetic Diseases, Inborn			
Metabolism, Inborn Errors			
Lipid Metabolism, Inborn Errors			
Lipidoses			
Sphingolipidoses			
Leukodystrophy, Globoid Cell	C16.320.565.398.641.803.585	C10.228.	C10.228.
		C10.228.	C10.228.
		C10.314.	C16.320.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Niemann-Pick Diseases	C16.320.565.398.641.803.730	C10.228.	C15.604.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Niemann-Pick Disease, Type A	C16.320.565.398.641.803.730.500	C10.228.	C15.604.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Niemann-Pick Disease, Type B	C16.320.565.398.641.803.730.750	C10.228.	C15.604.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Niemann-Pick Disease, Type C	C16.320.565.398.641.803.730.875	C10.228.	C15.604.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Sea-Blue Histiocyte Syndrome	C16.320.565.398.641.803.850	C10.228.	C15.604.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Sulfatidosis	C16.320.565.398.641.803.925	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
Leukodystrophy, Metachromatic	C16.320.565.398.641.803.925.500	C10.228.	C10.228.
		C10.228.	C10.228.
		C10.314.	C16.320.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Multiple Sulfatase Deficiency Disease	C16.320.565.398.641.803.925.750	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
Lipodystrophy, Congenital Generalized	C16.320.565.398.745	C17.800.	C18.452.
		C18.452.	C18.452.
Smith-Lemli-Opitz Syndrome	C16.320.565.398.850	C16.131.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Xanthomatosis, Cerebrotendinous	C16.320.565.398.925	C18.452.	C18.452.
Lysosomal Storage Diseases	C16.320.565.595	C18.452.	
Aspartylglucosaminuria	C16.320.565.595.100	C18.452.	
Cholesterol Ester Storage Disease	C16.320.565.595.201	C16.320.	C18.452.
		C18.452.	C18.452.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Metabolism, Inborn Errors
Lysosomal Storage Diseases
Cholesterol Ester Storage Disease
Wolman Disease

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. 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. C18.452.
Mucolipidoses	C16.320.565.595.554.590	C5.116. C16.320. C18.452. C18.452.	C10.228. C16.320. C18.452. 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. C18.452.
Fabry Disease	C16.320.565.595.554.825.200	C10.228. C14.907. C16.320. C18.452. C18.452. C18.452.	C10.228. C16.320. C16.320. C18.452. C18.452.
Farber Lipogranulomatosis	C16.320.565.595.554.825.250	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452. C18.452. C18.452.
Gangliosidoses	C16.320.565.595.554.825.300	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452. C18.452. C18.452.
Gangliosidoses, GM2	C16.320.565.595.554.825.300.300	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452. C18.452. 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.	C10.228. C16.320. C18.452. C18.452. C18.452. C18.452.
Tay-Sachs Disease	C16.320.565.595.554.825.300.300.840	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452. 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.	C16.320. C18.452. C18.452. C18.452.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Metabolism, Inborn Errors
Lysosomal Storage Diseases
Lysosomal Storage Diseases, Nervous System
Sphingolipidoses

Gangliosidosis, GM1	C16.320.565.595.554.825.300.400	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
Gaucher Disease	C16.320.565.595.554.825.400	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
Leukodystrophy, Globoid Cell	C16.320.565.595.554.825.590	C10.228.	C10.228.
		C10.228.	C10.228.
		C10.314.	C16.320.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Niemann-Pick Diseases	C16.320.565.595.554.825.700	C10.228.	C15.604.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Niemann-Pick Disease, Type A	C16.320.565.595.554.825.700.500	C10.228.	C15.604.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Niemann-Pick Disease, Type B	C16.320.565.595.554.825.700.750	C10.228.	C15.604.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Niemann-Pick Disease, Type C	C16.320.565.595.554.825.700.875	C10.228.	C15.604.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Sea-Blue Histiocyte Syndrome	C16.320.565.595.554.825.775	C10.228.	C15.604.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Sulfatidosis	C16.320.565.595.554.825.850	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
Leukodystrophy, Metachromatic	C16.320.565.595.554.825.850.500	C10.228.	C10.228.
		C10.228.	C10.228.
		C10.314.	C16.320.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Multiple Sulfatase Deficiency Disease	C16.320.565.595.554.825.850.750	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		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.	

Congenital, Hereditary, and Neonatal Diseases and Abnormalities			
Genetic Diseases, Inborn			
Metabolism, Inborn Errors			
Lysosomal Storage Diseases			
Mannosidase Deficiency Diseases			
beta-Mannosidosis			
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.
Pycnodysostosis	C16.320.565.595.800	C5.116. C18.452.	C16.320.
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.	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.	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. C18.452.
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.
Acatalasia	C16.320.565.663.25	C18.452.	

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Metabolism, Inborn Errors
Peroxisomal Disorders
Adrenoleukodystrophy

Adrenoleukodystrophy	C16.320.565.663.112	C10.228.	C10.228.
		C10.228.	C10.228.
		C10.314.	C10.597.
		C16.320.	C16.320.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	C19.53.
Chondrodysplasia Punctata, Rhizomelic	C16.320.565.663.200	C5.116.	C18.452.
Mevalonate Kinase Deficiency	C16.320.565.663.480	C10.228.	C15.378.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C20.683.
Refsum Disease	C16.320.565.663.760	C10.228.	C10.500.
		C10.574.	C10.668.
		C16.131.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
Refsum Disease, Infantile	C16.320.565.663.865	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Zellweger Syndrome	C16.320.565.663.970	C6.552.	C10.228.
		C12.777.	C13.351.
		C16.131.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Porphyrias	C16.320.565.708	C17.800.	C18.452.
		C18.452.	C18.452.
Porphyria, Erythropoietic	C16.320.565.708.250	C16.320.	C17.800.
		C17.800.	C18.452.
		C18.452.	C18.452.
Porphyrias, Hepatic	C16.320.565.708.400	C6.552.	C16.320.
		C17.800.	C17.800.
		C18.452.	C18.452.
		C18.452.	
Coproporphyria, Hereditary	C16.320.565.708.400.74	C6.552.	C16.320.
		C17.800.	C17.800.
		C18.452.	C18.452.
		C18.452.	
Porphyria, Acute Intermittent	C16.320.565.708.400.150	C6.552.	C16.320.
		C17.800.	C17.800.
		C18.452.	C18.452.
		C18.452.	
Porphyria Cutanea Tarda	C16.320.565.708.400.250	C6.552.	C16.320.
		C17.800.	C17.800.
		C18.452.	C18.452.
		C18.452.	
Porphyria, Hepatoerythropoietic	C16.320.565.708.400.437	C6.552.	C16.320.
		C17.800.	C17.800.
		C18.452.	C18.452.
		C18.452.	
Porphyria, Variegate	C16.320.565.708.400.625	C6.552.	C16.320.
		C17.800.	C17.800.
		C18.452.	C18.452.
		C18.452.	
Protoporphyria, Erythropoietic	C16.320.565.708.400.812	C6.552.	C16.320.
		C17.800.	C17.800.
		C18.452.	C18.452.
		C18.452.	
Progeria	C16.320.565.753	C18.452.	
Purine-Pyrimidine Metabolism, Inborn Errors	C16.320.565.798	C18.452.	

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Metabolism, Inborn Errors
Purine-Pyrimidine Metabolism, Inborn Errors
Dihydropyrimidine Dehydrogenase Deficiency

Dihydropyrimidine Dehydrogenase Deficiency	C16.320.565.798.183	C18.452.	
Gout	C16.320.565.798.368	C5.550.	C5.799.
		C18.452.	
Arthritis, Gouty	C16.320.565.798.368.410	C5.550.	C5.799.
		C18.452.	
Lesch-Nyhan Syndrome	C16.320.565.798.594	C10.228.	C10.574.
		C10.597.	C16.320.
		C16.320.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
Renal Tubular Transport, Inborn Errors	C16.320.565.861	C12.777.	C13.351.
		C18.452.	
Acidosis, Renal Tubular	C16.320.565.861.93	C12.777.	C13.351.
		C18.452.	C18.452.
Dent Disease	C16.320.565.861.271	C12.777.	C13.351.
		C16.320.	C18.452.
Fanconi Syndrome	C16.320.565.861.450	C12.777.	C12.777.
		C13.351.	C13.351.
		C18.452.	
Gitelman Syndrome	C16.320.565.861.491	C12.777.	C13.351.
		C18.452.	
Glycosuria, Renal	C16.320.565.861.532	C12.777.	C12.777.
		C13.351.	C13.351.
		C18.452.	C18.452.
Hypophosphatemia, Familial	C16.320.565.861.647	C12.777.	C13.351.
		C16.320.	C18.452.
		C18.452.	C18.452.
Hypophosphatemic Rickets, X-Linked Dominant	C16.320.565.861.647.500	C5.116.	C12.777.
		C13.351.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.654.	
Liddle Syndrome	C16.320.565.861.698	C12.777.	C13.351.
		C18.452.	
Oculocerebrorenal Syndrome	C16.320.565.861.750	C10.228.	C12.777.
		C13.351.	C16.131.
		C16.320.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Pseudohypoaldosteronism	C16.320.565.861.770	C12.777.	C13.351.
		C18.452.	
Renal Aminoacidurias	C16.320.565.861.885	C12.777.	C13.351.
		C18.452.	
Cystinuria	C16.320.565.861.885.250	C12.777.	C13.351.
		C18.452.	
Hartnup Disease	C16.320.565.861.885.457	C10.228.	C12.777.
		C13.351.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
Steroid Metabolism, Inborn Errors	C16.320.565.925	C18.452.	
Adrenal Hyperplasia, Congenital	C16.320.565.925.249	C12.706.	C13.351.
		C16.131.	C16.320.
		C18.452.	C19.53.
		C19.391.	
Antley-Bixler Syndrome Phenotype	C16.320.565.925.324	C5.116.	C5.660.
		C16.131.	C18.452.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Metabolism, Inborn Errors
Steroid Metabolism, Inborn Errors
Ichthyosis, X-Linked

Ichthyosis, X-Linked	C16.320.565.925.400	C16.131.	C16.320.
		C16.320.	C16.614.
		C17.800.	C17.800.
		C17.800.	C18.452.
Mineralocorticoid Excess Syndrome, Apparent	C16.320.565.925.500	C18.452.	
Smith-Lemli-Opitz Syndrome	C16.320.565.925.875	C16.131.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Muscular Dystrophies	C16.320.577	C5.651.	C10.668.
Distal Myopathies	C16.320.577.74	C5.651.	C10.668.
Glycogen Storage Disease Type VII	C16.320.577.149	C5.651.	C16.320.
		C18.452.	
Muscular Dystrophies, Limb-Girdle	C16.320.577.280	C5.651.	C10.668.
Sarcoglycanopathies	C16.320.577.280.500	C5.651.	C8.618.
		C10.668.	C14.280.
Muscular Dystrophy, Duchenne	C16.320.577.300	C5.651.	C10.668.
		C16.320.	
Muscular Dystrophy, Emery-Dreifuss	C16.320.577.350	C5.651.	C10.668.
		C16.320.	
Muscular Dystrophy, Facioscapulohumeral	C16.320.577.400	C5.651.	C10.668.
Muscular Dystrophy, Oculopharyngeal	C16.320.577.450	C5.651.	C10.668.
Myotonic Dystrophy	C16.320.577.500	C5.651.	C5.651.
		C10.574.	C10.668.
		C10.668.	C16.320.
Walker-Warburg Syndrome	C16.320.577.750	C10.500.	C11.270.
		C16.131.	
Myasthenic Syndromes, Congenital	C16.320.590	C10.668.	
Nail-Patella Syndrome	C16.320.600	C5.550.	C16.131.
		C17.800.	
Neoplastic Syndromes, Hereditary	C16.320.700	C4.700	
Adenomatous Polyposis Coli	C16.320.700.100	C4.557.	C4.588.
		C4.588.	C4.700.
		C6.301.	C6.405.
		C6.405.	C6.405.
		C6.405.	
Gardner Syndrome	C16.320.700.100.393	C4.557.	C4.588.
		C4.588.	C4.700.
		C6.301.	C6.405.
		C6.405.	C6.405.
		C6.405.	C16.131.
Basal Cell Nevus Syndrome	C16.320.700.175	C4.182.	C4.557.
		C4.557.	C4.700.
		C5.116.	C5.500.
		C7.320.	C16.131.
Birt-Hogg-Dube Syndrome	C16.320.700.212	C4.700.	
Colorectal Neoplasms, Hereditary Nonpolyposis	C16.320.700.250	C4.588.	C4.700.
		C6.301.	C6.405.
		C6.405.	C6.405.
		C18.452.	
Lynch Syndrome II	C16.320.700.250.500	C4.700.	
Muir-Torre Syndrome	C16.320.700.250.500.500	C4.588.	C4.700.
		C17.800.	C17.800.
		C17.800.	
Dysplastic Nevus Syndrome	C16.320.700.305	C4.557.	C4.700.
Exostoses, Multiple Hereditary	C16.320.700.330	C4.557.	C4.700.
		C5.116.	C5.116.
Hamartoma Syndrome, Multiple	C16.320.700.435	C4.445.	C4.651.
		C4.700.	

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Neoplastic Syndromes, Hereditary
Hereditary Breast and Ovarian Cancer Syndrome

Hereditary Breast and Ovarian Cancer Syndrome	C16.320.700.517	C4.588.	C4.588.
		C4.700.	C13.351.
		C13.351.	C17.800.
		C19.344.	C19.391.
Li-Fraumeni Syndrome	C16.320.700.600	C4.700.	C18.452.
Multiple Endocrine Neoplasia	C16.320.700.630	C4.588.	C4.651.
		C4.700.	C19.344.
Multiple Endocrine Neoplasia Type 1	C16.320.700.630.500	C4.588.	C4.651.
		C4.700.	C19.344.
Multiple Endocrine Neoplasia Type 2a	C16.320.700.630.505	C4.588.	C4.651.
		C4.700.	C19.344.
Multiple Endocrine Neoplasia Type 2b	C16.320.700.630.510	C4.588.	C4.651.
		C4.700.	C19.344.
Tuberous Sclerosis	C16.320.700.636	C4.445.	C4.651.
		C4.700.	C10.500.
		C10.562.	C10.574.
		C16.131.	C16.320.
Wilms Tumor	C16.320.700.642	C4.557.	C4.588.
		C4.700.	C12.758.
		C12.777.	C13.351.
		C13.351.	
Denys-Drash Syndrome	C16.320.700.642.220	C4.557.	C4.588.
		C4.700.	C12.706.
		C12.758.	C12.777.
		C13.351.	C13.351.
		C13.351.	C16.131.
		C19.391.	
WAGR Syndrome	C16.320.700.642.950	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.
		C19.391.	
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.	C4.700.
		C9.218.	C9.647.
		C10.292.	C10.292.
		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.	
Pelger-Huet Anomaly	C16.320.784	C15.378.	
Polycystic Kidney, Autosomal Recessive	C16.320.793	C12.777.	C13.351.
Pycnodysostosis	C16.320.812	C5.116.	C16.320.
		C18.452.	
Skin Diseases, Genetic	C16.320.850	C17.800.	
Albinism	C16.320.850.80	C11.270.	C16.320.
		C16.320.	C17.800.
		C17.800.	C18.452.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Skin Diseases, Genetic
Albinism
Albinism, Ocular

Albinism, Ocular	C16.320.850.80.90	C11.270. C16.320. C17.800. C18.452.	C16.320. C17.800.
Albinism, Oculocutaneous	C16.320.850.80.100	C11.270. C16.320. C17.800. C18.452.	C16.320. C17.800.
Hermanski-Pudlak Syndrome	C16.320.850.80.100.400	C11.270. C15.378. C15.378. C16.320. C17.800. C18.452.	C15.378. C15.378. C16.320. C17.800.
Piebaldism	C16.320.850.80.600	C16.320. C17.800. C18.452.	C16.320. C17.800.
Cutis Laxa	C16.320.850.180	C17.300.	C17.800.
Darier Disease	C16.320.850.190	C17.800.	C17.800.
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. C17.800.
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. C17.800.	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.
Erythrokeratodermia Variabilis	C16.320.850.337	C17.800. C17.800.	C17.800.
Hyalinosis, Systemic	C16.320.850.368		
Ichthyosiform Erythroderma, Congenital	C16.320.850.400	C16.131. C17.800. C17.800.	C16.614. C17.800.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Skin Diseases, Genetic
Ichthyosiform Erythroderma, Congenital
Hyperkeratosis, Epidermolytic

Hyperkeratosis, Epidermolytic	C16.320.850.400.375	C16.131. C17.800. C17.800.	C16.614. C17.800.
Ichthyosis, Lamellar	C16.320.850.400.410	C16.131. C17.800. C17.800.	C16.614. C17.800.
Netherton Syndrome	C16.320.850.400.705	C16.131. C16.320. C17.800. C17.800.	C16.131. C16.614. C17.800. C17.800.
Ichthyosis Bullosa of Siemens	C16.320.850.402	C16.131. C17.800. 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.
Papillon-Lefevre Disease	C16.320.850.475.600	C17.800.	C17.800.
Leukokeratosis, Hereditary Mucosal	C16.320.850.542	C17.800.	
Lipoid Proteinosis of Urbach and Wiethe	C16.320.850.595	C8.618.	
Monilethrix	C16.320.850.647	C16.131. C17.800.	C17.800.
Netherton Syndrome	C16.320.850.673	C16.131. C16.320. C17.800. C17.800.	C16.131. C16.614. C17.800. C17.800.
Pemphigus, Benign Familial	C16.320.850.700	C17.800.	C17.800.
Porokeratosis	C16.320.850.730	C17.800.	C17.800.
Porphyria, Erythropoietic	C16.320.850.738	C16.320. C17.800. C18.452.	C17.800. C18.452. C18.452.
Porphyrias, Hepatic	C16.320.850.742	C6.552. C17.800. C18.452. C18.452.	C16.320. C17.800. C18.452.
Coproporphyria, Hereditary	C16.320.850.742.74	C6.552. C17.800. C18.452. C18.452.	C16.320. C17.800. C18.452.
Porphyria, Acute Intermittent	C16.320.850.742.150	C6.552. C17.800. C18.452. C18.452.	C16.320. C17.800. C18.452.
Porphyria Cutanea Tarda	C16.320.850.742.250	C6.552. C17.800. C18.452. C18.452.	C16.320. C17.800. C18.452.
Porphyria, Hepatoerythropoietic	C16.320.850.742.437	C6.552. C17.800. C18.452. C18.452.	C16.320. C17.800. C18.452.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities			
Genetic Diseases, Inborn			
Skin Diseases, Genetic			
Porphyrias, Hepatic			
Porphyria, Variegate			
Porphyria, Variegate	C16.320.850.742.625	C6.552. C17.800. C18.452. C18.452.	C16.320. C17.800. C18.452.
Protoporphyria, Erythropoietic	C16.320.850.742.812	C6.552. C17.800. C18.452. C18.452.	C16.320. C17.800. C18.452.
Prolidase Deficiency	C16.320.850.746	C16.131. C16.320.	C16.131.
Pseudoxanthoma Elasticum	C16.320.850.750	C14.907. C16.131. C17.800.	C15.378. C17.300. C17.800.
Rothmund-Thomson Syndrome	C16.320.850.765	C16.131. C17.800. C18.452.	C16.614. C17.800.
Sjogren-Larsson Syndrome	C16.320.850.820	C16.131. C16.614. C17.800. C18.452.	C16.320. C17.800. C17.800.
Trichothiodystrophy Syndromes	C16.320.850.895	C16.131. C17.800.	C16.131. C17.800.
Xeroderma Pigmentosum	C16.320.850.970	C4.834. C17.800. C17.800. C18.452.	C16.131. C17.800. C17.800.
Weill-Marchesani Syndrome	C16.320.887	C5.116. C11.270. C16.320.	C5.116. C16.131. C17.300.
Werner Syndrome	C16.320.925	C18.452.	
Yellow Nail Syndrome	C16.320.962	C17.800. C23.300.	C17.800.
Infant, Newborn, Diseases	C16.614		
Amniotic Band Syndrome	C16.614.42		
Anemia, Neonatal	C16.614.53	C15.378.	
Fetofetal Transfusion	C16.614.53.344	C15.378.	
Fetomaternal Transfusion	C16.614.53.511	C15.378.	
Asphyxia Neonatorum	C16.614.92		
Birth Injuries	C16.614.131	C26.141	
Paralysis, Obstetric	C16.614.131.587	C26.141.	
Cystic Fibrosis	C16.614.213	C6.689. C16.320.	C8.381.
Epilepsy, Benign Neonatal	C16.614.258	C10.228.	
Erythroblastosis, Fetal	C16.614.304	C13.703. C16.300.	C15.378. C20.188.
Kernicterus	C16.614.304.502	C10.228. C18.452. C23.550.	C15.378. C20.188.
Hernia, Umbilical	C16.614.378	C23.300.	
Hydrophthalmos	C16.614.438	C11.250. C16.131.	C11.525.
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. C17.800. C17.800.	C16.320. C17.800.

Congenital, Hereditary, and Neonatal Diseases and Abnormalities			
Infant, Newborn, Diseases			
Ichthyosis			
Ichthyosiform Erythroderma, Congenital			
Hyperkeratosis, Epidermolytic			
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.	
Netherton Syndrome	C16.614.492.400.705	C16.131.	C16.131.
		C16.320.	C16.320.
		C17.800.	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.
Transient Tachypnea of the Newborn	C16.614.521.563.737	C8.381.	C8.618.
		C8.618.	C23.888.
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	C25.650	F3.900.
Nystagmus, Congenital	C16.614.643	C10.292.	C11.590.
Ophthalmia Neonatorum	C16.614.677	C1.252.	C1.252.
		C1.539.	C11.187.
		C11.294.	
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.	C16.320.
		C17.800.	C17.800.
		C18.452.	
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.	C5.116.
		C5.660.	C16.131.
		C16.320.	
Thrombocytopenia, Neonatal Alloimmune	C16.614.899	C15.378.	
Toxoplasmosis, Congenital	C16.614.909	C3.752.	C10.228.
Vitamin K Deficiency Bleeding	C16.614.940	C15.378.	C15.378.
		C18.654.	C18.654.
Wolman Disease	C16.614.947	C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	