

MeSH Tree Structures - 2015

C16 - DISEASES-CONGENITAL, HEREDITARY, 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.
Angelman Syndrome	C16.131.77.95	C16.320.	
		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, HEREDITARY, 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. C10.574. C10.597. C11.640. C12.777. C16.131. C16.320. C19.246.	C10.292. C10.597. C11.270. C11.966. C13.351. C16.320. C18.452. C19.700.
Donohue Syndrome	C16.131.77.313	C5.660. C18.452.	C16.320. C19.246.
Down Syndrome	C16.131.77.327	C10.597. C16.320.	C16.131.
Ectodermal Dysplasia	C16.131.77.350	C16.131. C17.800.	C16.320. C17.800.
Ectodermal Dysplasia 1, Anhidrotic	C16.131.77.350.198	C16.131. C16.320. C17.800.	C16.320. C17.800.
Ectodermal Dysplasia 3, Anhidrotic	C16.131.77.350.298	C16.131. C17.800.	C16.320. C17.800.
Ectodermal Dysplasia, Hypohidrotic, Autosomal Recessive	C16.131.77.350.348	C16.131. C17.800.	C16.320. C17.800.
Ellis-Van Creveld Syndrome	C16.131.77.350.398	C5.116. C16.320. C17.800.	C16.131. C17.800.
Focal Dermal Hypoplasia	C16.131.77.350.424	C5.116. C16.320. C17.800.	C16.131. C16.320. C17.800.
Neurocutaneous Syndromes	C16.131.77.350.712	C10.562 C16.320. C17.800.	C16.131. C17.800.
Fraser Syndrome	C16.131.77.371	C11.250.	C16.131.
Gardner Syndrome	C16.131.77.393	C4.557. C4.588. C6.301. C6.405. C6.405.	C4.588. C4.700. C6.405. C6.405. C16.320.
Heterotaxy Syndrome	C16.131.77.401	C14.240. C15.604.	C14.280. C16.131.
Holoprosencephaly	C16.131.77.410	C5.660. C16.131. C16.131.	C10.500. C16.131. C16.320.
Incontinentia Pigmenti	C16.131.77.445	C16.131. C17.800. C17.800.	C16.320. C17.800.
Isolated Noncompaction of the Ventricular Myocardium	C16.131.77.477	C14.240. C14.280. C16.320.	C14.280. C16.131.
Laurence-Moon Syndrome	C16.131.77.509	C10.228.	
LEOPARD Syndrome	C16.131.77.525	C5.660. C14.280. C16.131. C17.800.	C14.240. C14.280. C16.131.
Loeys-Dietz Syndrome	C16.131.77.537	C5.660. C14.907. C16.320.	C14.907. C14.907.
Marfan Syndrome	C16.131.77.550	C5.116. C14.280. C16.320.	C14.240. C16.131.
Mobius Syndrome	C16.131.77.578	C10.292.	C16.614.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

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. C17.800.	C16.320.
Netherton Syndrome	C16.131.77.619	C16.131. C16.320. C17.800. C17.800.	C16.320. C16.614. C17.800. C17.800.
Nevus, Sebaceous of Jadassohn	C16.131.77.633	C4.557.	C10.562.
Oculocerebrorenal Syndrome	C16.131.77.661	C10.228. C13.351. C16.320. C16.320. C18.452. C18.452.	C12.777. C16.320. C16.320. C18.452. C18.452.
Orofaciodigital Syndromes	C16.131.77.677	C5.116. C16.131. C16.320.	C5.660. C16.131.
Pallister-Hall Syndrome	C16.131.77.690	C4.445. C5.660. C10.228. C16.131.	C4.588. C10.228. C10.551.
Pentalogy of Cantrell	C16.131.77.696	C10.500.	C16.131.
POEMS Syndrome	C16.131.77.703	C10.668. C20.683.	C15.378.
Prader-Willi Syndrome	C16.131.77.730	C10.597. C16.320.	C16.131. C18.654.
Prolidase Deficiency	C16.131.77.735	C16.131. C16.320.	C16.320.
Proteus Syndrome	C16.131.77.740	C4.445. C5.116. C16.131.	C4.651. C5.660.
Prune Belly Syndrome	C16.131.77.745		
Rubella Syndrome, Congenital	C16.131.77.790	C2.782.	
Rubinstein-Taybi Syndrome	C16.131.77.804	C5.116. C10.597. C16.131.	C5.660. C16.131.
Short Rib-Polydactyly Syndrome	C16.131.77.850	C5.116. C16.131.	C5.660.
Silver-Russell Syndrome	C16.131.77.855	C5.660. C16.320.	C16.131. C16.320.
Smith-Lemli-Opitz Syndrome	C16.131.77.860	C16.320. C18.452. C18.452.	C16.320. C16.320. C18.452.
Smith-Magenis Syndrome	C16.131.77.879	C10.281. C16.320.	C16.131.
Sotos Syndrome	C16.131.77.889	C16.131.	C16.320.
Trichothiodystrophy Syndromes	C16.131.77.899	C16.131. C17.800.	C16.320. C17.800.
Waardenburg Syndrome	C16.131.77.938		
Weill-Marchesani Syndrome	C16.131.77.941	C5.116. C11.270. C16.320.	C5.116. C16.320. C17.300.
Wolf-Hirschhorn Syndrome	C16.131.77.944	C16.131.	C16.320.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

**Congenital Abnormalities
Abnormalities, Multiple
Wolfram Syndrome**

Wolfram Syndrome	C16.131.77.951	C9.218. C10.574. C10.597. C11.640. C12.777. C16.131. C16.320. C19.246.	C10.292. C10.597. C11.270. C11.966. C13.351. C16.320. C18.452. C19.700.
Zellweger Syndrome	C16.131.77.970	C6.552. C12.777. C16.320. C18.452. C18.452.	C10.228. C13.351. C16.320. C18.452.
Abnormalities, Radiation-Induced Abnormalities, Severe Teratoid Anencephaly Twins, Conjoined Aicardi Syndrome	C16.131.80 C16.131.85 C16.131.85.197 C16.131.85.806 C16.131.162	C26.733. C10.500.	N6.850. C16.131.
Cardiovascular Abnormalities Heart Defects, Congenital 22q11 Deletion Syndrome	C16.131.240 C16.131.240.400 C16.131.240.400.21	C14.240. C14.240. C5.660. C14.280. C16.131. C16.131. C16.320.	C14.280. C14.240. C14.240. C15.604. C16.131. C16.131. C19.642.
DiGeorge Syndrome	C16.131.240.400.21.500	C5.660. C14.280. C16.131. C16.131. C16.320.	C14.240. C15.604. C16.131. C16.131. C19.642.
Alagille Syndrome	C16.131.240.400.44	C6.130. C14.240. C16.320.	C6.552. C16.131.
Aortic Coarctation Arrhythmogenic Right Ventricular Dysplasia	C16.131.240.400.90 C16.131.240.400.145	C14.240. C14.240. C14.280.	C14.280. C14.280.
Barth Syndrome	C16.131.240.400.172	C14.240. C16.131. C16.320.	C14.280. C16.320. C18.452.
Cor Triatriatum Coronary Vessel Anomalies Bland White Garland Syndrome	C16.131.240.400.200 C16.131.240.400.210 C16.131.240.400.210.249	C14.240. C14.240. C14.240. C14.280. C16.131.	C14.280. C14.280. C14.240. C14.907. C23.300.
Myocardial Bridging Crisscross Heart Dextrocardia	C16.131.240.400.210.500 C16.131.240.400.220 C16.131.240.400.280	C14.240. C14.240. C14.240. C16.131.	C14.280. C14.280. C14.280.
Kartagener Syndrome	C16.131.240.400.280.500	C8.127. C8.695. C14.240. C16.131. C16.320.	C8.200. C9.150. C14.280. C16.131.
Ductus Arteriosus, Patent Ebstein Anomaly Ectopia Cordis Eisenmenger Complex Heart Septal Defects	C16.131.240.400.340 C16.131.240.400.395 C16.131.240.400.422 C16.131.240.400.450 C16.131.240.400.560	C14.240. C14.240. C14.240. C14.240. C14.240.	C14.280. C14.280. C14.280. C14.280.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Cardiovascular Abnormalities

Heart Defects, Congenital

Heart Septal Defects

Aortopulmonary Septal Defect

Aortopulmonary Septal Defect

Truncus Arteriosus, Persistent

Endocardial Cushion Defects

Heart Septal Defects, Atrial

Foramen Ovale, Patent

Lutembacher Syndrome

Heart Septal Defects, Ventricular

Heterotaxy Syndrome

Hypoplastic Left Heart Syndrome

Isolated Noncompaction of the Ventricular Myocardium

LEOPARD Syndrome

Levocardia

Long QT Syndrome

Andersen Syndrome

Jervell-Lange Nielsen Syndrome

Romano-Ward Syndrome

Marfan Syndrome

Noonan Syndrome

Tetralogy of Fallot

Transposition of Great Vessels

Double Outlet Right Ventricle

Tricuspid Atresia

Trilogy of Fallot

Turner Syndrome

Wolff-Parkinson-White Syndrome

Vascular Malformations

Arterio-Arterial Fistula

Bland White Garland Syndrome

Arteriovenous Malformations

Arteriovenous Fistula

Intracranial Arteriovenous Malformations

Vein of Galen Malformations

	C16.131.240.400.560.98	C14.240.	C14.280.
	C16.131.240.400.560.98.500	C14.240.	C14.280.
	C16.131.240.400.560.350	C14.240.	C14.280.
	C16.131.240.400.560.375	C14.240.	C14.280.
	C16.131.240.400.560.375.258	C14.240.	C14.280.
	C16.131.240.400.560.375.518	C14.240.	C14.280.
	C16.131.240.400.560.540	C14.240.	C14.280.
	C16.131.240.400.592	C14.240.	C14.280.
		C15.604.	C16.131.
	C16.131.240.400.625	C14.240.	C14.280.
	C16.131.240.400.655	C14.240.	C14.280.
		C14.280.	C16.131.
		C16.320.	
	C16.131.240.400.685	C5.660.	C14.240.
		C14.280.	C14.280.
		C16.131.	C16.131.
		C17.800.	
	C16.131.240.400.701	C14.240.	C14.280.
		C16.131.	
	C16.131.240.400.715	C14.280.	C23.550.
	C16.131.240.400.715.70	C14.280.	C23.550.
	C16.131.240.400.715.440	C14.280.	
	C16.131.240.400.715.720	C14.280.	C23.550.
	C16.131.240.400.720	C5.116.	C14.240.
		C14.280.	C16.131.
		C16.320.	C17.300.
	C16.131.240.400.784	C5.660.	C14.240.
		C14.280.	C16.131.
		C17.300.	
	C16.131.240.400.849	C14.240.	C14.280.
	C16.131.240.400.915	C14.240.	C14.280.
	C16.131.240.400.915.300	C14.240.	C14.280.
	C16.131.240.400.920	C14.240.	C14.280.
		C14.280.	
	C16.131.240.400.960	C14.240.	C14.280.
	C16.131.240.400.970	C12.706.	C12.706.
		C13.351.	C13.351.
		C14.240.	C14.280.
		C16.131.	C16.131.
		C16.131.	C16.320.
		C19.391.	C19.391.
	C16.131.240.400.980	C14.280.	
	C16.131.240.850	C14.240.	
	C16.131.240.850.500	C14.240.	C14.907.
		C23.300.	
	C16.131.240.850.500.500	C14.240.	C14.240.
		C14.280.	C14.907.
		C16.131.	C23.300.
	C16.131.240.850.750	C14.240.	C14.907.
	C16.131.240.850.750.125	C14.240.	C14.240.
		C14.907.	C14.907.
		C23.300.	
	C16.131.240.850.750.295	C10.228.	C10.500.
		C14.240.	C14.240.
		C14.907.	C14.907.
		C16.131.	C16.131.
	C16.131.240.850.750.295.500	C10.228.	C10.500.
		C14.240.	C14.240.
		C14.907.	C14.907.
		C16.131.	C16.131.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Cardiovascular Abnormalities

Vascular Malformations

Central Nervous System Vascular Malformations

Central Nervous System Vascular Malformations

Intracranial Arteriovenous Malformations

Vein of Galen Malformations

May-Thurner Syndrome

Pulmonary Atresia

Scimitar Syndrome

Single Umbilical Artery

Telangiectasia, Hereditary Hemorrhagic

Chromosome Disorders

22q11 Deletion Syndrome

DiGeorge Syndrome

Angelman Syndrome

Beckwith-Wiedemann Syndrome

Branchio-Oto-Renal Syndrome

Cri-du-Chat Syndrome

De Lange Syndrome

Down Syndrome

Holoprosencephaly

Jacobsen Distal 11q Deletion Syndrome

Prader-Willi Syndrome

Rubinstein-Taybi Syndrome

Sex Chromosome Disorders

Fragile X Syndrome

Orofaciodigital Syndromes

Sex Chromosome Disorders of Sex Development

C16.131.240.850.875	C10.500. C16.131.	C14.240.
C16.131.240.850.875.500	C10.228. C14.240. C14.907.	C10.500. C14.240. C14.907.
C16.131.240.850.875.500.500	C16.131. C10.228. C14.240. C14.907. C16.131.	C10.500. C14.240. C14.907. C14.907. C16.131.
C16.131.240.850.890	C14.240.	C14.907.
C16.131.240.850.906	C14.240.	C14.280.
C16.131.240.850.937	C8.381. C14.240. C16.131.	C8.695. C14.907.
C16.131.240.850.952	C14.240.	
C16.131.240.850.968	C14.907.	C14.907.
C16.131.260	C15.378. C16.320.	
C16.131.260.19	C5.660. C14.280.	C14.240. C15.604.
C16.131.260.19.500	C16.131. C16.131. C16.320. C19.642. C5.660. C14.280.	C16.131. C16.131. C16.131. C19.642. C14.240. C15.604.
C16.131.260.40	C16.131. C16.131. C16.320. C10.228. C16.320.	C16.131. C16.131. C19.642. C16.131.
C16.131.260.80	C16.131.	C16.320.
C16.131.260.90	C16.131.	C16.320.
C16.131.260.190	C10.597. C16.320.	C16.131.
C16.131.260.210	C10.597. C16.320.	C16.131.
C16.131.260.260	C10.597. C16.320.	C16.131.
C16.131.260.380	C5.660. C16.131. C16.131.	C10.500. C16.131. C16.320.
C16.131.260.440	C15.378.	C16.320.
C16.131.260.700	C10.597. C16.320.	C16.131. C18.654.
C16.131.260.790	C5.116. C10.597. C16.131.	C5.660. C16.131. C16.320.
C16.131.260.830	C16.320.	
C16.131.260.830.300	C10.597. C16.320.	C16.320. C16.320.
C16.131.260.830.670	C5.116. C16.131. C16.320.	C5.660. C16.131. C16.320.
C16.131.260.830.835	C12.706. C16.131.	C13.351. C16.320.
	C19.391.	

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Chromosome Disorders

Sex Chromosome Disorders

Sex Chromosome Disorders of Sex Development

Gonadal Dysgenesis, Mixed

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.

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.
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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.
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Congenital Microtia

C16.131.287	C9.218.	
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Digestive System Abnormalities

Anus, Imperforate

C16.131.314	C6.198	
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Biliary Atresia

C16.131.314.94	C6.198.	
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Choledochal Cyst

C16.131.314.125	C6.130.	C6.198.
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Caroli Disease

C16.131.314.184	C4.182.	C6.130.
	C6.198.	

Diaphragmatic Eventration

C16.131.314.184.500	C6.130.	C6.198.
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Esophageal Atresia

C16.131.314.244	C6.198.	
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Hirschsprung Disease

C16.131.314.330	C6.198.	C6.405.
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Intestinal Atresia

C16.131.314.439	C6.198.	C6.405.
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Meckel Diverticulum

C16.131.314.466	C6.198.	C6.405.
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C16.131.314.556	A3.556.	A3.556.
	C6.198.	C23.300.

Eye Abnormalities

Aniridia

C16.131.384	C11.250	
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WAGR Syndrome

C16.131.384.79	C11.250.	C11.270.
	C11.941.	C16.320.

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.	
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Blepharophimosis

C16.131.384.190	C11.250.	C11.338.
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C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Eye Abnormalities

Coloboma

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.
Hernias, Diaphragmatic, Congenital	C16.131.433	C23.300.	
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	
Arthrogyrosis	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.
Holoprosencephaly	C16.131.621.207.410	C5.660.	C10.500.
		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.	

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Musculoskeletal Abnormalities

Craniofacial Abnormalities

Megalencephaly

Megalencephaly	C16.131.621.207.532	C5.660. C16.131.	C10.500.
Hemimegalencephaly	C16.131.621.207.532.500	C5.660. C16.131.	C10.500.
Maxillofacial Abnormalities	C16.131.621.207.540	C5.660. C16.131.	C7.650.
Cherubism	C16.131.621.207.540.170	C5.116. C7.320.	C5.500. C16.320.
Dentofacial Deformities	C16.131.621.207.540.315	C5.660. C16.131.	C7.650.
Jaw Abnormalities	C16.131.621.207.540.460	C5.500. C7.320. C16.131.	C5.660. C7.650.
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.
Retrognathia	C16.131.621.207.540.460.827	C5.500. C7.320. C7.650.	C5.660. C7.320. C16.131.
Microcephaly	C16.131.621.207.620	C5.660. C16.131.	C10.500.
Porencephaly	C16.131.621.207.620.500	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. 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.
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.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Musculoskeletal Abnormalities

Hip Dislocation, Congenital

Hip Dislocation, Congenital

Klippel-Feil Syndrome

Laryngomalacia

C16.131.621.449

C16.131.621.551

C16.131.621.568

C5.660.

C5.116.

C5.182.

C9.400.

C5.660.

C8.360.

C17.300.

Limb Deformities, Congenital

Arachnodactyly

Brachydactyly

Ectromelia

Foot Deformities, Congenital

Clubfoot

Hand Deformities, Congenital

Lower Extremity Deformities, Congenital

Polydactyly

Pallister-Hall Syndrome

C16.131.621.585

C16.131.621.585.174

C16.131.621.585.262

C16.131.621.585.350

C16.131.621.585.380

C16.131.621.585.380.500

C16.131.621.585.425

C16.131.621.585.512

C16.131.621.585.600

C16.131.621.585.600.374

C5.660.

C5.660.

C5.660.

C5.660.

C5.330.

C5.330.

C5.390.

C5.660.

C5.660.

C4.445.

C5.660.

C10.228.

C10.228.

C16.131.

C5.116.

C16.131.

C4.445.

C5.116.

C16.131.

C5.116.

C5.660.

C5.660.

C5.660.

C5.660.

C16.131.

C16.131.

C5.116.

C5.660.

C5.116.

C5.660.

C16.614.

C5.660.

C5.116.

C5.660.

C5.660.

C16.320.

C5.116.

C5.660.

C5.660.

C16.131.

C5.660.

C5.660.

C16.131.

C16.131.

C16.131.

C5.116.

C5.660.

C5.660.

C5.660.

C16.131.

C16.131.

C5.116.

C5.660.

Short Rib-Polydactyly Syndrome

C16.131.621.585.600.750

Proteus Syndrome

C16.131.621.585.620

Syndactyly

C16.131.621.585.800

Acrocephalosyndactyly

C16.131.621.585.800.100

Poland Syndrome

C16.131.621.585.800.756

Thanatophoric Dysplasia

C16.131.621.585.984

Upper Extremity Deformities, Congenital

Pectus Carinatum

C16.131.621.585.988

C16.131.621.745

Synostosis

Antley-Bixler Syndrome Phenotype

C16.131.621.906

C16.131.621.906.181

Craniosynostoses

C16.131.621.906.364

Acrocephalosyndactyly

C16.131.621.906.364.100

Syndactyly

C16.131.621.906.819

Acrocephalosyndactyly

C16.131.621.906.819.100

Poland Syndrome

C16.131.621.906.819.756

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Musculoskeletal Abnormalities

Tracheobronchomalacia

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.
 Holoprosencephaly	C16.131.666.34.875	C5.660. C16.131. C16.131.	C10.500. C16.131. C16.320.
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.	C10.500. C10.668. C16.320. C18.452.
 Spastic Paraplegia, Hereditary	C16.131.666.300.820	C10.500. C10.668.	C10.574. C16.320.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Nervous System Malformations

Hereditary Sensory and Autonomic Neuropathies

Hereditary Sensory and Autonomic Neuropathies

C16.131.666.310	C10.114.	C10.314.
	C10.500.	C10.574.
	C10.668.	C10.668.
	C16.320.	C20.111.
	C10.114.	C10.177.
	C10.314.	C10.500.
	C10.574.	C10.668.
	C10.668.	C16.320.
	C20.111.	

Dysautonomia, Familial

C16.131.666.310.309	C10.114.	C10.177.
	C10.314.	C10.500.
	C10.574.	C10.668.
	C10.668.	C16.320.
	C20.111.	

Hydranencephaly

C16.131.666.450	C10.500.	
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Malformations of Cortical Development

C16.131.666.507	C10.500.	
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Malformations of Cortical Development, Group I

C16.131.666.507.400	C10.500.	
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Megalencephaly

C16.131.666.507.400.249	C5.660.	C10.500.
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Hemimegalencephaly

C16.131.666.507.400.249.500	C16.131.	
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	C5.660.	C10.500.
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Microcephaly

C16.131.666.507.400.500	C16.131.	
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	C5.660.	C10.500.
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Tuberous Sclerosis

C16.131.666.507.400.750	C16.131.	
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	C4.445.	C4.651.
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	C4.700.	C10.500.
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	C10.562.	C10.574.
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	C16.320.	C16.320.
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Malformations of Cortical Development, Group II

C16.131.666.507.450	C10.500.	
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Classical Lissencephalies and Subcortical Band

Heterotopias

C16.131.666.507.450.230	C10.500.	C10.500.
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	C16.131.	C16.320.
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Cobblestone Lissencephaly

C16.131.666.507.450.249	C10.500.	C10.500.
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	C16.131.	
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Lissencephaly

C16.131.666.507.450.499	C10.500.	
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Classical Lissencephalies and Subcortical Band

Heterotopias

C16.131.666.507.450.499.230	C10.500.	C10.500.
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	C16.131.	C16.320.
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Cobblestone Lissencephaly

C16.131.666.507.450.499.249	C10.500.	C10.500.
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	C16.131.	
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Walker-Warburg Syndrome

C16.131.666.507.450.499.249.500	C10.500.	C11.270.
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	C16.320.	
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Periventricular Nodular Heterotopia

C16.131.666.507.450.750	C10.500.	
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Malformations of Cortical Development, Group III

C16.131.666.507.500	C10.500.	
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Polymicrogyria

C16.131.666.507.500.500	C10.500.	
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Porencephaly

C16.131.666.507.500.625	C5.660.	C10.500.
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	C16.131.	
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Schizencephaly

C16.131.666.507.500.750	C10.500.	
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Neural Tube Defects

C16.131.666.680	C10.500.	
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Anencephaly

C16.131.666.680.196	C10.500.	C16.131.
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Arnold-Chiari Malformation

C16.131.666.680.291	C10.500.	
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Encephalocele

C16.131.666.680.488	C10.500.	C23.300.
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Meningocele

C16.131.666.680.598	C10.500.	C23.300.
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Meningomyelocele

C16.131.666.680.610	C10.500.	
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Pentalogy of Cantrell

C16.131.666.680.705	C10.500.	C16.131.
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Spinal Dysraphism

C16.131.666.680.800	C10.500.	
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Spina Bifida Cystica

C16.131.666.680.800.730	C10.500.	
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Spina Bifida Occulta

C16.131.666.680.800.750	C10.500.	
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Septo-Optic Dysplasia

C16.131.666.845	C10.500.	
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Respiratory System Abnormalities

C16.131.740	C8.695	
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Bronchogenic Cyst

C16.131.740.195	C4.182.	C8.127.
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	C8.695.	
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Bronchopulmonary Sequestration

C16.131.740.214	C8.695.	
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Choanal Atresia

C16.131.740.271	C8.460.	C8.695.
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	C9.603.	
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C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Respiratory System Abnormalities

Cystic Adenomatoid Malformation of Lung, Congenital

Cystic Adenomatoid Malformation of Lung, Congenital	C16.131.740.290	C8.381.	C8.695.
Kartagener Syndrome	C16.131.740.501	C8.127.	C8.200.
		C8.695.	C9.150.
		C14.240.	C14.280.
		C16.131.	C16.131.
		C16.320.	
Laryngocele	C16.131.740.650	C8.360.	C8.695.
		C9.400.	
Laryngostenosis	C16.131.740.658	C8.360.	C9.400.
Scimitar Syndrome	C16.131.740.815	C8.381.	C8.695.
		C14.240.	C14.907.
		C16.131.	
Tracheobronchomegaly	C16.131.740.830	C8.127.	C8.695.
		C8.907.	
Situs Inversus	C16.131.810		
Dextrocardia	C16.131.810.250	C14.240.	C14.280.
		C16.131.	
Kartagener Syndrome	C16.131.810.250.500	C8.127.	C8.200.
		C8.695.	C9.150.
		C14.240.	C14.280.
		C16.131.	C16.131.
		C16.320.	
Levocardia	C16.131.810.700	C14.240.	C14.280.
		C16.131.	
Skin Abnormalities	C16.131.831	C17.800.	
Acrodermatitis	C16.131.831.66	C17.800.	C17.800.
Carney Complex	C16.131.831.108	C4.557.	C4.588.
		C14.280.	C16.131.
Dyskeratosis Congenita	C16.131.831.150	C16.320.	C16.320.
		C17.800.	C17.800.
Ectodermal Dysplasia	C16.131.831.350	C16.131.	C16.320.
		C17.800.	C17.800.
Ectodermal Dysplasia 1, Anhidrotic	C16.131.831.350.198	C16.131.	C16.320.
		C16.320.	C17.800.
		C17.800.	
Ectodermal Dysplasia 3, Anhidrotic	C16.131.831.350.298	C16.131.	C16.320.
		C17.800.	C17.800.
Ectodermal Dysplasia, Hypohidrotic, Autosomal Recessive	C16.131.831.350.348	C16.131.	C16.320.
		C17.800.	C17.800.
Ellis-Van Creveld Syndrome	C16.131.831.350.398	C5.116.	C16.131.
		C16.320.	C17.800.
		C17.800.	
Focal Dermal Hypoplasia	C16.131.831.350.424	C5.116.	C16.131.
		C16.320.	C16.320.
		C17.800.	C17.800.
Neurocutaneous Syndromes	C16.131.831.350.712	C10.562	C16.131.
		C16.320.	C17.800.
		C17.800.	
Pachyonychia Congenita	C16.131.831.350.856	C16.320.	C17.800.
		C17.800.	C17.800.
Steatocystoma Multiplex	C16.131.831.350.856.500	C16.320.	C17.800.
		C17.800.	C17.800.
Ehlers-Danlos Syndrome	C16.131.831.428	C14.907.	C15.378.
		C16.320.	C17.300.
		C17.800.	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.	

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Skin Abnormalities

Epidermolysis Bullosa

Epidermolysis Bullosa Dystrophica

Epidermolysis Bullosa Dystrophica

C16.131.831.493.160

C16.320.
C17.300.
C17.800.
C17.800.

Epidermolysis Bullosa, Junctional

C16.131.831.493.170

C16.320.
C17.800.

Epidermolysis Bullosa Simplex

C16.131.831.493.180

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

Ichthyosis

C16.131.831.512

C16.614.
C17.800.

Ichthyosiform Erythroderma, Congenital

C16.131.831.512.400

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

Hyperkeratosis, Epidermolytic

C16.131.831.512.400.375

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

Ichthyosis, Lamellar

C16.131.831.512.400.410

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

Netherton Syndrome

C16.131.831.512.400.705

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

Ichthyosis Bullosa of Siemens

C16.131.831.512.408

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

Ichthyosis Vulgaris

C16.131.831.512.410

C16.320.
C17.800.

Ichthyosis, X-Linked

C16.131.831.512.420

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

Sjogren-Larsson Syndrome

C16.131.831.512.723

C16.320.
C16.320.
C16.614.
C17.800.
C17.800.

Incontinentia Pigmenti

C16.131.831.580

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

Port-Wine Stain

C16.131.831.675

C17.800.

Prolidase Deficiency

C16.131.831.720

C16.131.
C16.320.

Pseudoxanthoma Elasticum

C16.131.831.766

C14.907.
C15.378.
C16.320.
C17.300.

Rothmund-Thomson Syndrome

C16.131.831.775

C17.800.
C16.320.
C16.614.
C17.800.
C17.800.

Sclerema Neonatorum

C16.131.831.812

C18.452.
C16.614.

Trichothiodystrophy Syndromes

C16.131.831.874

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

Xeroderma Pigmentosum

C16.131.831.936

C17.800.
C4.834.
C16.320.
C17.800.
C17.800.

Stomatognathic System Abnormalities

Maxillofacial Abnormalities

Dentofacial Deformities

C16.131.850

C7.650

C16.131.850.500

C5.660.
C7.650.

C16.131.850.500.229

C16.131.
C5.660.
C7.650.
C16.131.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Stomatognathic System Abnormalities

Maxillofacial Abnormalities

Jaw Abnormalities

Jaw Abnormalities

C16.131.850.500.460

C5.500.
C7.320. C5.660.
C7.650.

Cleft Palate

C16.131.850.500.460.185

C16.131.
C5.500. C5.660.
C7.320. C7.465.
C7.650. C7.650.

Micrognathism

C16.131.850.500.460.457

C16.131. C16.131.
C5.500. C5.660.
C7.320. C7.650.

Pierre Robin Syndrome

C16.131.850.500.460.606

C16.131.
C5.500. C5.660.
C7.320. C7.650.

Prognathism

C16.131.850.500.460.655

C16.131.
C5.500. C5.500.
C5.660. C7.320.
C7.320. C7.650.

Retrognathia

C16.131.850.500.460.827

C16.131.
C5.500. C5.660.
C7.320. C7.320.
C7.650. C16.131.

Mouth Abnormalities

Cleft Lip

C16.131.850.525

C7.465. C7.650.
C7.465. C7.650.

Cleft Palate

C16.131.850.525.185

C7.650. C5.660.
C7.320. C7.465.
C7.650. C7.650.

Fibromatosis, Gingival

C16.131.850.525.304

C16.131. C16.131.
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.

46, XX Testicular Disorders of Sex Development

C16.131.939.316.64.124

C19.391. F3.800.
C12.706. C13.351.
C19.391.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Urogenital Abnormalities

Disorders of Sex Development

46, XX Disorders of Sex Development

Gonadal Dysgenesis, 46,XX

Gonadal Dysgenesis, 46,XX

C16.131.939.316.64.249

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

C12.706.
C13.351.
C19.391.

Hyperandrogenism

C16.131.939.316.64.500

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

C12.706.
C13.351.
C19.391.

46, XY Disorders of Sex Development

C16.131.939.316.96

C12.706.
C19.391.

C13.351.
F3.800.

Androgen-Insensitivity Syndrome

C16.131.939.316.96.500

C12.706.
C16.320.

C13.351.
C19.391.

Denys-Drash Syndrome

C16.131.939.316.96.562

C4.557.
C4.700.
C12.758.
C13.351.
C13.351.
C19.391.

C4.588.
C12.706.
C12.777.
C13.351.
C16.320.

Frasier Syndrome

C16.131.939.316.96.624

C12.706.
C13.351.

C12.777.
C13.351.

Gonadal Dysgenesis, 46,XY

C16.131.939.316.96.687

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

C19.391.
C12.706.
C13.351.

Gonadoblastoma

C16.131.939.316.96.687.500

C4.557.
C12.706.
C13.351.
C16.131.

C4.557.
C12.706.
C13.351.
C19.391.

Kallmann Syndrome

C16.131.939.316.96.750

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

C13.351.
C19.391.

WAGR Syndrome

C16.131.939.316.96.875

C19.391.
C4.557.
C4.700.
C11.250.
C11.941.
C12.758.
C13.351.
C13.351.
C16.131.
C16.131.
C16.320.
C19.391.

C4.588.
C10.597.
C11.270.
C12.706.
C12.777.
C13.351.
C16.131.
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.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Congenital Abnormalities

Urogenital Abnormalities

Disorders of Sex Development

Gonadal Dysgenesis

Gonadal Dysgenesis, 46,XY

Gonadal Dysgenesis, 46,XY

C16.131.939.316.309.388
C12.706. C12.706.
C13.351. C13.351.
C16.131. C19.391.
C19.391.

Gonadoblastoma

C16.131.939.316.309.388.500
C4.557. C4.557.
C12.706. C12.706.
C13.351. C13.351.
C16.131. C19.391.
C19.391.

Gonadal Dysgenesis, Mixed

C16.131.939.316.309.391
C12.706. C12.706.
C13.351. C13.351.
C16.131. C16.131.
C16.320. C19.391.
C19.391.

Sexual Infantilism

C16.131.939.316.309.631
C12.706. C13.351.
C19.391. C19.391.

Turner Syndrome

C16.131.939.316.309.872
C12.706. C12.706.
C13.351. C13.351.
C14.240. C14.280.
C16.131. C16.131.
C16.131. C16.320.
C19.391. C19.391.

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

Retrocaval Ureter

C16.131.939.831
C13.703. C16.300.
C16.131.939.915
C12.706. C13.351.

Fetal Diseases

Chorioamnionitis

C16.300
C13.703. C13.703.
C16.300.30
C13.703. C13.703.

Echogenic Bowel

Erythroblastosis, Fetal

C16.300.50
C13.703.
C16.300.60
C13.703. C15.378.
C16.614. C20.306

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Genetic Diseases, Inborn
Ataxia Telangiectasia

Ataxia Telangiectasia	C16.320.80	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.
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.
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.
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.
Hermanski-Pudlak Syndrome	C16.320.99.515	C11.270. C15.378. C15.378. C16.320. C17.800.	C15.378. C16.320. C15.378. C16.320. C17.800.
Hypoprothrombinemias	C16.320.99.550	C18.452. 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.
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. C20.673.	C15.378. C16.320. C20.673.
Brugada Syndrome	C16.320.100	C14.280.	
CADASIL	C16.320.129	C10.228. C10.228. C14.907.	C10.228. C10.228. C14.907.
Camurati-Engelmann Syndrome	C16.320.144	C5.116.	
Cardiomyopathy, Hypertrophic, Familial	C16.320.160	C14.280.	C14.280.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

CHARGE Syndrome

**CHARGE Syndrome
Cherubism**

C16.320.165 C16.131.
C16.320.170 C5.116. C5.500.
C7.320. C16.131.

**Chromosome Disorders
22q11 Deletion Syndrome**

C16.320.180 C16.131.
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
Branchio-Oto-Renal Syndrome
Cri-du-Chat Syndrome**

C16.320.180.80 C16.131. C16.131.
C16.320.180.90 C16.131. C16.131.
C16.320.180.190 C10.597. C16.131.
C16.131.

De Lange Syndrome

C16.320.180.210 C10.597. C16.131.
C16.131.

Down Syndrome

C16.320.180.260 C10.597. C16.131.
C16.131.

Holoprosencephaly

C16.320.180.380 C5.660. C10.500.
C16.131. C16.131.
C16.131. C16.131.

**Jacobsen Distal 11q Deletion Syndrome
Prader-Willi Syndrome**

C16.320.180.440 C15.378. C16.131.
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
Fragile X Syndrome**

C16.320.180.830 C16.131.
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.

Silver-Russell Syndrome

C16.320.180.870 C19.391. C19.391.
C5.660. C16.131.

Smith-Magenis Syndrome

C16.320.180.887 C16.131. C16.320.
C10.281. C16.131.
C16.131.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Chromosome Disorders

Sotos Syndrome

**Sotos Syndrome
WAGR Syndrome**

C16.320.180.905
C16.320.180.940

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

Thanatophoric Dysplasia

C16.320.240.500.500

C5.116. C5.116.
C5.660. C16.131.
C16.614.

Cockayne Syndrome

C16.320.240.562

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

Congenital Hypothyroidism

C16.320.240.625

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

Laron Syndrome

C16.320.240.750

C5.116. C19.297.

Mulibrey Nanism

C16.320.240.875

C5.116.

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. C11.270.
C16.131. C16.131.
C16.320.

Albinism

C16.320.290.40

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

Albinism, Ocular

C16.320.290.40.90

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

Albinism, Oculocutaneous

C16.320.290.40.100

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

Hermanski-Pudlak Syndrome

C16.320.290.40.100.400

C11.270. C15.378.
C15.378. C15.378.

Piebaldism

C16.320.290.40.600

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

Aniridia

C16.320.290.78

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

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Eye Diseases, Hereditary

Aniridia

WAGR Syndrome

WAGR Syndrome

C16.320.290.78.950

C4.557. C4.588.
C4.700. C10.597.
C11.250. C11.270.
C11.941. C12.706.
C12.758. C12.777.
C13.351. C13.351.
C13.351. C16.131.
C16.131. C16.131.
C16.320. C16.320.
C19.391.

Choroideremia

C16.320.290.142

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

Corneal Dystrophies, Hereditary

C16.320.290.162

C11.204. C11.270.

Corneal Dystrophy, Juvenile Epithelial of Meesmann

C16.320.290.162.204

C11.204. C11.270.

Fuchs' Endothelial Dystrophy

C16.320.290.162.410

C11.204. C11.270.

Duane Retraction Syndrome

C16.320.290.235

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

Gyrate Atrophy

C16.320.290.468

C11.270. C11.941.

Optic Atrophies, Hereditary

C16.320.290.564

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

Optic Atrophy, Autosomal Dominant

C16.320.290.564.100

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

Optic Atrophy, Hereditary, Leber

C16.320.290.564.400

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

Wolfram Syndrome

C16.320.290.564.980

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

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.

Frasier Syndrome

C16.320.306

C16.320. C17.300.
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.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Genetic Diseases, X-Linked

Barth Syndrome

Barth Syndrome	C16.320.322.68	C14.240. C16.131. C16.320.	C14.280. C16.131. 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. C16.320.	C11.941.
Dent Disease	C16.320.322.100	C12.777. C16.320.	C13.351. C18.452.
Dyskeratosis Congenita	C16.320.322.108	C16.131. C17.800.	C16.320. C17.800.
Ectodermal Dysplasia 1, Anhidrotic	C16.320.322.116	C16.131. C16.320. C17.800.	C16.131. C17.800.
Fabry Disease	C16.320.322.124	C10.228. C14.907. C16.320. C18.452. C18.452. C18.452.	C10.228. C16.320. C16.320. C18.452. C18.452.
Focal Dermal Hypoplasia	C16.320.322.186	C5.116. C16.131. C17.800.	C16.131. C16.320. C17.800.
Glycogen Storage Disease Type IIb	C16.320.322.201	C10.597. C16.320.	C14.280. C18.452.
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.
Hyper-IgM Immunodeficiency Syndrome, Type 1	C16.320.322.237	C15.378.	C20.673.
Ichthyosis, X-Linked	C16.320.322.241	C16.131. C16.320. C17.800. C17.800.	C16.320. C16.614. C17.800. C18.452.
Isolated Noncompaction of the Ventricular Myocardium	C16.320.322.370	C14.240. C14.280. C16.131.	C14.280. C16.131.
Mental Retardation, X-Linked	C16.320.322.500	C10.597.	C16.320.
Adrenoleukodystrophy	C16.320.322.500.124	C10.228. C10.228. C10.597. C16.320. C16.320. C18.452. C18.452. C18.452. C19.53.	C10.228. C10.314. C16.320. C16.320. C18.452. C18.452. C18.452.
Classical Lissencephalies and Subcortical Band Heterotopias	C16.320.322.500.186	C10.500. C16.131.	C10.500. C16.131.
Coffin-Lowry Syndrome	C16.320.322.500.249	C10.597.	C16.320.
Fragile X Syndrome	C16.320.322.500.500	C10.597. C16.320.	C16.131. C16.320.
Lesch-Nyhan Syndrome	C16.320.322.500.625	C10.228. C10.597. C16.320. C16.320. C18.452.	C10.574. C16.320. C16.320. C18.452.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Genetic Diseases, X-Linked

Mental Retardation, X-Linked

Menkes Kinky Hair Syndrome

Menkes Kinky Hair Syndrome

C16.320.322.500.687

C10.228.
C10.597.
C16.320.
C16.320.
C17.800.
C18.452.
C18.452.

C10.574.
C16.320.
C16.320.
C17.800.
C18.452.

Mucopolysaccharidosis II

C16.320.322.500.750

C10.597.
C16.320.
C17.300.
C18.452.

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

Pyruvate Dehydrogenase Complex Deficiency Disease

C16.320.322.500.875

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

C10.597.
C16.320.
C18.452.
C18.452.

Rett Syndrome

C16.320.322.500.937

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

C10.597.
C16.320.
C10.668.

Muscular Dystrophy, Duchenne

C16.320.322.562

C5.651.
C16.320.

C10.668.

Muscular Dystrophy, Emery-Dreifuss

C16.320.322.625

C5.651.
C16.320.

C10.668.

Oculocerebrorenal Syndrome

C16.320.322.750

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

C12.777.
C16.131.
C16.320.
C18.452.
C18.452.

Ornithine Carbamoyltransferase Deficiency Disease

C16.320.322.828

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

C16.320.
C18.452.

Pelizaeus-Merzbacher Disease

C16.320.322.906

C10.228.
C10.314.
C18.452.

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

Wiskott-Aldrich Syndrome

C16.320.322.937

C15.378.
C15.378.
C20.673.

C15.378.
C16.320.
C20.673.

X-Linked Combined Immunodeficiency Diseases

C16.320.322.968

C16.614.

C20.673.

Genetic Diseases, Y-Linked

Hajdu-Cheney Syndrome

C16.320.338

C16.320.355

C5.116.
C16.131.

C5.116.

Hemoglobinopathies

Anemia, Sickle Cell

C16.320.365

C16.320.365.155

C15.378.
C15.378.
C16.320.

C15.378.
C15.378.

Acute Chest Syndrome

C16.320.365.155.219

C8.381.
C15.378.

C8.618.
C15.378.

Hemoglobin SC Disease

C16.320.365.155.440

C16.320.
C15.378.

C15.378.
C15.378.

Sickle Cell Trait

C16.320.365.155.668

C15.378.
C16.320.

C15.378.
C15.378.

Hemoglobin C Disease

C16.320.365.463

C15.378.
C16.320.

C15.378.
C15.378.

Thalassemia

C16.320.365.826

C15.378.
C16.320.

C15.378.
C15.378.

alpha-Thalassemia

C16.320.365.826.100

C15.378.
C16.320.

C15.378.
C15.378.

Hydrops Fetalis

C16.320.365.826.100.350

C13.703.
C15.378.
C20.306.

C15.378.
C16.300.
C23.888.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Hemoglobinopathies

Thalassemia

beta-Thalassemia

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.

Hereditary Degenerative Disorders, Nervous System

C16.320.400

C10.574.

Alexander Disease

C16.320.400.24

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.

Canavan Disease

C16.320.400.150

C18.452.

C18.452.

C10.228.

C10.228.

C10.314.

C10.574.

C16.320.

C18.452.

C18.452.

Cockayne Syndrome

C16.320.400.200

C5.116.

C10.574.

C16.131.

C16.320.

C18.452.

Dystonia Musculorum Deformans

C16.320.400.330

C10.228.

C10.228.

C10.574.

Gerstmann-Straussler-Scheinker Disease

C16.320.400.350

C10.228.

C10.574.

C10.574.

Hepatolenticular Degeneration

C16.320.400.361

C6.552.

C10.228.

C10.228.

C10.228.

C10.574.

C16.320.

C16.320.

C18.452.

C18.452.

C18.452.

Hereditary Central Nervous System Demyelinating Diseases

C16.320.400.367

C10.228.

C10.228.

C10.314.

C10.574.

C16.320.

C18.452.

C18.452.

Hereditary Sensory and Motor Neuropathy

C16.320.400.375

C10.500.

C10.574.

Alstrom Syndrome

C16.320.400.375.99

C10.668.

C16.131.

C10.500.

C10.574.

C10.668.

C11.270.

C16.131.

C16.131.

C16.320.

Charcot-Marie-Tooth Disease

C16.320.400.375.200

C10.500.

C10.574.

C10.668.

C16.131.

Giant Axonal Neuropathy

C16.320.400.375.490

C10.500.

C10.574.

C10.668.

C10.668.

C16.131.

Refsum Disease

C16.320.400.375.780

C10.228.

C10.500.

C10.574.

C10.668.

C16.131.

C16.320.

C16.320.

C18.452.

C18.452.

C18.452.

Spastic Paraplegia, Hereditary

C16.320.400.375.820

C10.500.

C10.574.

C10.668.

C16.131.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

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

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

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Hereditary Degenerative Disorders, Nervous System

Myotonic Dystrophy

Myotonic Dystrophy	C16.320.400.542	C5.651. C10.574. C10.668.	C5.651. C10.668. C16.320.
Neuroacanthocytosis	C16.320.400.550	C10.228.	
Neurofibromatoses	C16.320.400.560	C4.557. C10.562. C16.320.	C4.700. C10.574.
Neurofibromatosis 1	C16.320.400.560.400	C4.557. C10.562. C10.668.	C4.700. C10.574. C16.320.
Neurofibromatosis 2	C16.320.400.560.700	C4.557. C4.557. C9.218. C10.292. C10.562. C16.320.	C4.557. C4.700. C9.647. C10.292. C10.574.
Neuronal Ceroid-Lipofuscinoses	C16.320.400.600	C10.574. C18.452.	C16.320. C18.452.
Optic Atrophies, Hereditary	C16.320.400.630	C10.292. C11.270. C16.320.	C10.574. C11.640.
Optic Atrophy, Autosomal Dominant	C16.320.400.630.100	C10.292. C11.270. C16.320.	C10.574. C11.640. C18.452.
Optic Atrophy, Hereditary, Leber	C16.320.400.630.400	C10.292. C11.270. C16.320.	C10.574. C11.640. C18.452.
Wolfram Syndrome	C16.320.400.630.980	C9.218. C10.574. C10.597. C11.640. C12.777. C16.131. C16.320. C19.246.	C10.292. C10.597. C11.270. C11.966. C13.351. C16.131. C18.452. C19.700.
Pantothenate Kinase-Associated Neurodegeneration	C16.320.400.650	C10.228. C10.228.	C10.228. C10.228.
Rett Syndrome	C16.320.400.700	C10.574. C16.320.	C10.597. C16.320.
Spinal Muscular Atrophies of Childhood	C16.320.400.765	C10.228. C10.574. C10.668.	C10.574. C10.668.
Spinocerebellar Degenerations	C16.320.400.780	C10.228. C10.574.	C10.228.
Friedreich Ataxia	C16.320.400.780.200	C10.228. C10.574.	C10.228. C18.452.
Myoclonic Cerebellar Dyssynergia	C16.320.400.780.500	C10.228. C10.574.	C10.228.
Olivopontocerebellar Atrophies	C16.320.400.780.750	C10.228. C10.228. C10.574. C10.574.	C10.228. C10.228. C10.574.
Spinocerebellar Ataxias	C16.320.400.780.875	C10.228. C10.228. C10.597.	C10.228. C10.574.
Machado-Joseph Disease	C16.320.400.780.875.500	C10.228. C10.228. C10.597.	C10.228. C10.574.
Tourette Syndrome	C16.320.400.820	C10.228. C10.574.	C10.228. F3.550.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Hereditary Degenerative Disorders, Nervous System

Tuberous Sclerosis

Tuberous Sclerosis	C16.320.400.880	C4.445. C4.700. C10.562. C16.131.	C4.651. C10.500. C10.574. C16.320.
Unverricht-Lundborg Syndrome	C16.320.400.940	C10.228.	C10.574.
Hyper-IgM Immunodeficiency Syndrome	C16.320.413	C15.378.	C20.673.
Hyperthyroxinemia, Familial Dysalbuminemic	C16.320.427	C19.874.	
Kallmann Syndrome	C16.320.467	C12.706. C16.131. C19.391.	C13.351. C19.391.
Kartagener Syndrome	C16.320.480	C8.127. C8.695. C14.240. C16.131. C16.131.	C8.200. C9.150. C14.280. C16.131.
Lennox Gastaut Syndrome	C16.320.495	C10.228.	
Loeys-Dietz Syndrome	C16.320.510	C5.660. C14.907. C16.131.	C14.907. C14.907.
Marfan Syndrome	C16.320.540	C5.116. C14.280. C16.131.	C14.240. 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. C17.800.	C16.320. C17.800. C18.452.
Albinism, Ocular	C16.320.565.100.102.90	C11.270. C16.320. C17.800.	C16.320. C17.800. C18.452.
Albinism, Oculocutaneous	C16.320.565.100.102.100	C11.270. C16.320. C17.800. C18.452.	C16.320. C17.800. C18.452. C18.452.
Hermanski-Pudlak Syndrome	C16.320.565.100.102.100.400	C11.270. C15.378. C15.378. C16.320. C17.800. C18.452.	C15.378. C15.378. C16.320. C16.320. C17.800.
Piebaldism	C16.320.565.100.102.600	C16.320. C17.800. C18.452.	C16.320. C17.800.
Alkaptonuria	C16.320.565.100.187	C18.452.	
Hyperglycinemia, Nonketotic	C16.320.565.100.477	C10.228. C18.452. C18.452.	C16.320. C18.452.
Hyperhomocysteinemia	C16.320.565.100.480	C18.452. C18.654.	C18.452.
Homocystinuria	C16.320.565.100.480.500	C10.228. C17.300. C18.452.	C16.320. C18.452.
Hyperlysinemias	C16.320.565.100.544	C10.228. C18.452. C18.452.	C16.320. C18.452.
Maple Syrup Urine Disease	C16.320.565.100.608	C10.228. C18.452. C18.452.	C16.320. 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.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Amino Acid Metabolism, Inborn Errors

Multiple Carboxylase Deficiency

Biotinidase Deficiency

Biotinidase Deficiency

C16.320.565.100.620.100 C16.320. C18.452.

Holocarboxylase Synthetase Deficiency

C16.320.565.100.620.380 C16.320. C18.452.

Phenylketonurias

C16.320.565.100.766 C10.228. C16.320.
C18.452. C18.452.

Phenylketonuria, Maternal

C16.320.565.100.766.500 C10.228. C13.703.
C16.320. 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.

Urea Cycle Disorders, Inborn

C16.320.565.100.940 C10.228. C16.320.
C18.452. C18.452.

Argininosuccinic Aciduria

C16.320.565.100.940.124 C10.228. C16.320.
C18.452. C18.452.

Carbamoyl-Phosphate Synthase I Deficiency Disease

C16.320.565.100.940.249 C10.228. C16.320.
C18.452. C18.452.

Citrullinemia

C16.320.565.100.940.374 C10.228. C16.320.
C18.452. C18.452.

Hyperargininemia

C16.320.565.100.940.500 C10.228. C16.320.
C18.452. C18.452.

Ornithine Carbamoyltransferase Deficiency Disease

C16.320.565.100.940.750 C10.228. C16.320.
C16.320. C18.452.

Amino Acid Transport Disorders, Inborn

Hartnup Disease

C16.320.565.151 C18.452.
C16.320.565.151.355 C10.228. C12.777.
C13.351. C16.320.

Oculocerebrorenal Syndrome

C16.320.565.151.600 C10.228. C12.777.
C13.351. C16.131.

Amyloidosis, Familial

Amyloid Neuropathies, Familial

C16.320.565.176 C18.452. C18.452.
C16.320.565.176.50 C10.574. C10.668.
C16.320. C18.452.

Cerebral Amyloid Angiopathy, Familial

C16.320.565.176.160 C10.228. C10.228.
C10.228. C14.907. C14.907.
C18.452. C18.452.

Brain Diseases, Metabolic, Inborn

C16.320.565.189 C18.452. C18.452.
C10.228. C18.452.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Brain Diseases, Metabolic, Inborn

Cerebral Amyloid Angiopathy, Familial

Cerebral Amyloid Angiopathy, Familial	C16.320.565.189.168	C10.228. C10.228. C14.907. C16.320. 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.
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.
Hereditary Central Nervous System Demyelinating Diseases	C16.320.565.189.362	C10.228. C10.314. C16.320. C18.452.	C10.228. C10.574. C18.452.
Adrenoleukodystrophy	C16.320.565.189.362.250	C10.228. C10.228. C10.597. C16.320. C16.320. C18.452. C18.452.	C10.228. C10.314. C16.320. C16.320. C18.452. C18.452.
Alexander Disease	C16.320.565.189.362.312	C10.228. C10.314. C16.320. C18.452.	C10.228. C10.574. C18.452.
Canavan Disease	C16.320.565.189.362.375	C10.228. C10.314. C16.320. C18.452.	C10.228. C10.574. C18.452.
Leukodystrophy, Globoid Cell	C16.320.565.189.362.500	C10.228. C10.228. C16.320. C16.320. C18.452. C18.452. C18.452.	C10.228. C10.314. C16.320. C18.452. C18.452.
Leukodystrophy, Metachromatic	C16.320.565.189.362.550	C10.228. C10.228. C16.320. C18.452. C18.452. C18.452.	C10.228. C10.314. C16.320. C18.452. C18.452.
Pelizaeus-Merzbacher Disease	C16.320.565.189.362.775	C10.228. C10.314. C18.452.	C10.228. C16.320. C18.452.
Homocystinuria	C16.320.565.189.365	C10.228. C17.300. C18.452.	C16.320. C18.452.
Hyperglycinemia, Nonketotic	C16.320.565.189.375	C10.228. C18.452. C18.452.	C16.320. C18.452.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Brain Diseases, Metabolic, Inborn

Hyperlysinemias

Hyperlysinemias

C16.320.565.189.380

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

C16.320.
C18.452.

Leigh Disease

C16.320.565.189.412

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

C16.320.
C18.452.

Lesch-Nyhan Syndrome

C16.320.565.189.425

C10.228.
C10.597.
C16.320.
C16.320.

C10.574.
C16.320.

Lysosomal Storage Diseases, Nervous System

C16.320.565.189.435

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

C16.320.
C18.452.

Fucosidosis

C16.320.565.189.435.295

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

C16.320.
C18.452.

Glycogen Storage Disease Type II

C16.320.565.189.435.340

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

C16.320.
C18.452.

Mucopolidoses

C16.320.565.189.435.590

C5.116.
C16.320.
C18.452.
C18.452.

C10.228.
C16.320.

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.

Fabry Disease

C16.320.565.189.435.825.200

C10.228.
C14.907.
C16.320.
C18.452.

C10.228.
C16.320.

Farber Lipogranulomatosis

C16.320.565.189.435.825.250

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

C16.320.
C18.452.

Gangliosidoses

C16.320.565.189.435.825.300

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

C16.320.
C18.452.

Gangliosidoses, GM2

C16.320.565.189.435.825.300.300

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

C16.320.
C18.452.

Sandhoff Disease

C16.320.565.189.435.825.300.300.249

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

C10.228.
C16.320.

Tay-Sachs Disease

C16.320.565.189.435.825.300.300.500

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

C16.320.
C18.452.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Brain Diseases, Metabolic, Inborn

Lysosomal Storage Diseases, Nervous System

Sphingolipidoses

Tay-Sachs Disease, AB Variant	C16.320.565.189.435.825.300.300.750	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452.
Gangliosidosis, GM1	C16.320.565.189.435.825.300.400	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452.
Sandhoff Disease	C16.320.565.189.435.825.300.700	C10.228. C16.320. C18.452. C18.452.	C10.228. C16.320.
Gaucher Disease	C16.320.565.189.435.825.400	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452.
Leukodystrophy, Globoid Cell	C16.320.565.189.435.825.590	C10.228. C10.228. C16.320. C16.320. C18.452. C18.452.	C10.228. C10.314. C16.320. C18.452.
Niemann-Pick Diseases	C16.320.565.189.435.825.700	C10.228. C16.320. C18.452. C18.452.	C15.604. C16.320. C18.452.
Niemann-Pick Disease, Type A	C16.320.565.189.435.825.700.500	C10.228. C16.320. C18.452. C18.452.	C15.604. C16.320. C18.452.
Niemann-Pick Disease, Type B	C16.320.565.189.435.825.700.750	C10.228. C16.320. C18.452. C18.452.	C15.604. C16.320. C18.452.
Niemann-Pick Disease, Type C	C16.320.565.189.435.825.700.875	C10.228. C16.320. C18.452. C18.452.	C15.604. C16.320. C18.452.
Sea-Blue Histiocyte Syndrome	C16.320.565.189.435.825.775	C10.228. C16.320. C18.452. C18.452.	C15.604. C16.320. C18.452.
Sulfatidosis	C16.320.565.189.435.825.850	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452.
Leukodystrophy, Metachromatic	C16.320.565.189.435.825.850.500	C10.228. C10.228. C16.320. C16.320. C18.452. C18.452.	C10.228. C10.314. C16.320. C18.452. C18.452.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Brain Diseases, Metabolic, Inborn

Lysosomal Storage Diseases, Nervous System

Sphingolipidoses

Multiple Sulfatase Deficiency Disease	C16.320.565.189.435.825.850.750	C10.228. C16.320. C18.452. C18.452. C18.452.	C16.320. C18.452. C18.452.
Maple Syrup Urine Disease	C16.320.565.189.520	C10.228. C18.452. C18.452.	C16.320. C18.452.
MELAS Syndrome	C16.320.565.189.535	C5.651. C10.228. C14.907. C18.452.	C10.228. C10.668. C18.452. C18.452.
Menkes Kinky Hair Syndrome	C16.320.565.189.540	C10.228. C10.597. C16.320. C16.320. C18.452. C18.452.	C10.574. C16.320. C16.320. C17.800. C18.452.
MERRF Syndrome	C16.320.565.189.545	C5.651. C10.228. C18.452. C18.452.	C10.228. C10.668. C18.452.
Oculocerebrorenal Syndrome	C16.320.565.189.640	C10.228. C13.351. C16.320. C16.320. C18.452. C18.452.	C12.777. C16.131. C16.320. C18.452. C18.452.
Peroxisomal Disorders	C16.320.565.189.680	C10.228. C18.452. C18.452.	C16.320. C18.452.
Adrenoleukodystrophy	C16.320.565.189.680.100	C10.228. C10.228. C10.597. C16.320. C16.320. C18.452. C18.452. C18.452. C19.53.	C10.228. C10.314. C16.320. C16.320. C18.452. C18.452. C18.452.
Mevalonate Kinase Deficiency	C16.320.565.189.680.430	C10.228. C16.320. C18.452. C18.452.	C15.378. C16.320. C18.452.
Refsum Disease	C16.320.565.189.680.760	C10.228. C10.574. C16.131. C16.320. C18.452.	C10.500. C10.668. C16.320. C18.452.
Refsum Disease, Infantile	C16.320.565.189.680.865	C10.228. C18.452. C18.452.	C16.320. C18.452.
Zellweger Syndrome	C16.320.565.189.680.970	C6.552. C12.777. C16.131. C18.452. C18.452.	C10.228. C13.351. C16.320. C18.452.
Phenylketonurias	C16.320.565.189.687	C10.228. C18.452. C18.452.	C16.320. C18.452.
Phenylketonuria, Maternal	C16.320.565.189.687.500	C10.228. C16.320. C18.452.	C13.703. C18.452. C18.452.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Brain Diseases, Metabolic, Inborn

Pyruvate Carboxylase Deficiency Disease

Pyruvate Carboxylase Deficiency Disease

C16.320.565.189.725

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

Pyruvate Dehydrogenase Complex Deficiency Disease

C16.320.565.189.750

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

Tyrosinemias

C16.320.565.189.875

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

Urea Cycle Disorders, Inborn

C16.320.565.189.937

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

Argininosuccinic Aciduria

C16.320.565.189.937.124

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

Carbamoyl-Phosphate Synthase I Deficiency Disease

C16.320.565.189.937.249

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

Citrullinemia

C16.320.565.189.937.374

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

Hyperargininemia

C16.320.565.189.937.500

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

Ornithine Carbamoyltransferase Deficiency Disease

C16.320.565.189.937.750

C10.228.
C16.320.
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.

Galactosemias

C16.320.565.202.355

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

Glucosephosphate Dehydrogenase Deficiency

C16.320.565.202.402

C15.378.
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.

Glycogen Storage Disease Type IIb

C16.320.565.202.449.510

C10.597.
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.
C16.320.
C18.452.

Glycogen Storage Disease Type VIII

C16.320.565.202.449.620

C16.320.
C18.452.

Hyperoxaluria, Primary

C16.320.565.202.460

C12.777.
C13.351.
C18.452.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Carbohydrate Metabolism, Inborn Errors

Lactose Intolerance

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.

Mucopolidoses

C16.320.565.202.670

C5.116.
C16.320.

C10.228.
C16.320.

Mucopolysaccharidoses

C16.320.565.202.715

C16.320.
C18.452.

C17.300.
C18.452.

Mucopolysaccharidosis I

C16.320.565.202.715.640

C16.320.
C18.452.

C17.300.
C18.452.

Mucopolysaccharidosis II

C16.320.565.202.715.645

C10.597.
C16.320.

C16.320.
C18.452.

Mucopolysaccharidosis III

C16.320.565.202.715.650

C16.320.
C18.452.

C17.300.
C18.452.

Mucopolysaccharidosis IV

C16.320.565.202.715.655

C16.320.
C18.452.

C17.300.
C18.452.

Mucopolysaccharidosis VI

C16.320.565.202.715.670

C16.320.
C18.452.

C17.300.
C18.452.

Mucopolysaccharidosis VII

C16.320.565.202.715.675

C16.320.
C18.452.

C17.300.
C18.452.

Multiple Carboxylase Deficiency

C16.320.565.202.720

C16.320.
C18.452.

C18.452.

Biotinidase Deficiency

C16.320.565.202.720.100

C16.320.
C18.452.

C18.452.

Holocarboxylase Synthetase Deficiency

C16.320.565.202.720.380

C16.320.
C18.452.

C18.452.

Pyruvate Metabolism, Inborn Errors

C16.320.565.202.810

C18.452.

Leigh Disease

C16.320.565.202.810.444

C10.228.
C18.452.

C16.320.
C18.452.

Pyruvate Carboxylase Deficiency Disease

C16.320.565.202.810.666

C10.228.
C18.452.

C16.320.
C18.452.

Pyruvate Dehydrogenase Complex Deficiency Disease

C16.320.565.202.810.766

C10.228.
C16.320.

C10.597.
C16.320.

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.

Barth Syndrome

C16.320.565.398.224

C14.240.

C14.280.
C16.131.

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.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Lipid Metabolism, Inborn Errors

Hyperlipoproteinemia Type IV

Hyperlipoproteinemia Type IV

C16.320.565.398.487

C18.452.

C18.452.

Hyperlipoproteinemia Type V

C16.320.565.398.493

C18.452.

C18.452.

Hypolipoproteinemias

C16.320.565.398.500

C18.452.

C18.452.

Hypoalphalipoproteinemias

C16.320.565.398.500.330

C18.452.

C18.452.

Lecithin Acyltransferase Deficiency

C16.320.565.398.500.330.500

C18.452.

C18.452.

Tangier Disease

C16.320.565.398.500.330.750

C10.668.

C18.452.

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.

Wolman Disease

C16.320.565.398.641.201.500

C16.320.

C16.614.

Neuronal Ceroid-Lipofuscinoses

C16.320.565.398.641.509

C10.574.

C16.320.

Sjogren-Larsson Syndrome

C16.320.565.398.641.723

C18.452.

C18.452.

Sphingolipidoses

C16.320.565.398.641.803

C10.228.

C16.320.

Fabry Disease

C16.320.565.398.641.803.300

C16.320.

C18.452.

C18.452.

C18.452.

C10.228.

C10.228.

C14.907.

C16.320.

C16.320.

C16.320.

C18.452.

C18.452.

C18.452.

C18.452.

C18.452.

C18.452.

Farber Lipogranulomatosis

C16.320.565.398.641.803.325

C18.452.

C18.452.

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.

Gangliosidoses, GM2

C16.320.565.398.641.803.350.300

C16.320.

C18.452.

C18.452.

C18.452.

C18.452.

C18.452.

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

C18.452.

C18.452.

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

C18.452.

C18.452.

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

C18.452.

C18.452.

C10.228.

C16.320.

C16.320.

C18.452.

C18.452.

C18.452.

C18.452.

C18.452.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Lipid Metabolism, Inborn Errors

Lipidoses

Sphingolipidoses

Gangliosidosis, GM1	C16.320.565.398.641.803.350.360	C10.228. C16.320. C18.452. C18.452. C18.452.	C16.320. C18.452. C18.452.
Gaucher Disease	C16.320.565.398.641.803.441	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452. C18.452.
Leukodystrophy, Globoid Cell	C16.320.565.398.641.803.585	C10.228. C10.228. C16.320. C16.320. C18.452. C18.452.	C10.228. C10.314. C16.320. C18.452. C18.452.
Niemann-Pick Diseases	C16.320.565.398.641.803.730	C18.452. C10.228. C16.320. C18.452. C18.452.	C15.604. C16.320. C18.452. C18.452.
Niemann-Pick Disease, Type A	C16.320.565.398.641.803.730.500	C18.452. C10.228. C16.320. C18.452. C18.452.	C15.604. C16.320. C18.452. C18.452.
Niemann-Pick Disease, Type B	C16.320.565.398.641.803.730.750	C18.452. C10.228. C16.320. C18.452. C18.452.	C15.604. C16.320. C18.452. C18.452.
Niemann-Pick Disease, Type C	C16.320.565.398.641.803.730.875	C18.452. C10.228. C16.320. C18.452. C18.452.	C15.604. C16.320. C18.452. C18.452.
Sea-Blue Histiocyte Syndrome	C16.320.565.398.641.803.850	C18.452. C10.228. C16.320. C18.452. C18.452.	C15.604. C16.320. C18.452. C18.452.
Sulfatidosis	C16.320.565.398.641.803.925	C18.452. C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452. C18.452.
Leukodystrophy, Metachromatic	C16.320.565.398.641.803.925.500	C18.452. C10.228. C10.228. C16.320. C16.320. C18.452. C18.452. C18.452.	C18.452. C10.228. C10.314. C16.320. C18.452. C18.452. C18.452.
Multiple Sulfatase Deficiency Disease	C16.320.565.398.641.803.925.750	C18.452. C10.228. C16.320. C18.452. C18.452.	C18.452. C16.320. C18.452. C18.452.
Lipodystrophy, Congenital Generalized	C16.320.565.398.745	C18.452. C17.800.	C18.452. C18.452.
Smith-Lemli-Opitz Syndrome	C16.320.565.398.850	C18.452. C16.131. C18.452. C18.452.	C16.320. C16.320. C18.452.
Xanthomatosis, Cerebrotendinous	C16.320.565.398.925	C18.452.	C18.452.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Lysosomal Storage Diseases

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.
Wolman Disease	C16.320.565.595.201.500	C18.452.	C18.452.
		C16.320.	C16.614.
		C18.452.	C18.452.
		C18.452.	
Cystinosis	C16.320.565.595.377	C18.452.	
Lysosomal Storage Diseases, Nervous System	C16.320.565.595.554	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Fucosidosis	C16.320.565.595.554.295	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Glycogen Storage Disease Type II	C16.320.565.595.554.340	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Mucopolidoses	C16.320.565.595.554.590	C5.116.	C10.228.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
Sialic Acid Storage Disease	C16.320.565.595.554.810	C10.228.	C16.320.
		C18.452.	C18.452.
		C18.452.	
Sphingolipidoses	C16.320.565.595.554.825	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
Fabry Disease	C16.320.565.595.554.825.200	C10.228.	C10.228.
		C14.907.	C16.320.
		C16.320.	C16.320.
		C18.452.	C18.452.
		C18.452.	C18.452.
		C18.452.	
Farber Lipogranulomatosis	C16.320.565.595.554.825.250	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
Gangliosidoses	C16.320.565.595.554.825.300	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
Gangliosidoses, GM2	C16.320.565.595.554.825.300.300	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
Sandhoff Disease	C16.320.565.595.554.825.300.300.800	C10.228.	C10.228.
		C16.320.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.
Tay-Sachs Disease	C16.320.565.595.554.825.300.300.840	C10.228.	C16.320.
		C16.320.	C18.452.
		C18.452.	C18.452.
		C18.452.	C18.452.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Lysosomal Storage Diseases

Lysosomal Storage Diseases, Nervous System

Sphingolipidoses

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.
Gangliosidosis, GM1	C16.320.565.595.554.825.300.400	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452. C18.452.
Gaucher Disease	C16.320.565.595.554.825.400	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452. C18.452.
Leukodystrophy, Globoid Cell	C16.320.565.595.554.825.590	C10.228. C10.228. C16.320. C16.320. C18.452. C18.452. C18.452.	C10.228. C10.314. C16.320. C18.452. C18.452.
Niemann-Pick Diseases	C16.320.565.595.554.825.700	C10.228. C16.320. C18.452. C18.452.	C15.604. C16.320. C18.452.
Niemann-Pick Disease, Type A	C16.320.565.595.554.825.700.500	C10.228. C16.320. C18.452. C18.452.	C15.604. C16.320. C18.452.
Niemann-Pick Disease, Type B	C16.320.565.595.554.825.700.750	C10.228. C16.320. C18.452. C18.452.	C15.604. C16.320. C18.452.
Niemann-Pick Disease, Type C	C16.320.565.595.554.825.700.875	C10.228. C16.320. C18.452. C18.452.	C15.604. C16.320. C18.452.
Sea-Blue Histiocyte Syndrome	C16.320.565.595.554.825.775	C10.228. C16.320. C18.452. C18.452.	C15.604. C16.320. C18.452.
Sulfatidosis	C16.320.565.595.554.825.850	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452.
Leukodystrophy, Metachromatic	C16.320.565.595.554.825.850.500	C10.228. C10.228. C16.320. C16.320. C18.452. C18.452. C18.452.	C10.228. C10.314. C16.320. C18.452.
Multiple Sulfatase Deficiency Disease	C16.320.565.595.554.825.850.750	C10.228. C16.320. C18.452. C18.452.	C16.320. C18.452.
Mannosidase Deficiency Diseases	C16.320.565.595.577	C16.320. C18.452.	C18.452.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Metabolism, Inborn Errors

Lysosomal Storage Diseases

Mannosidase Deficiency Diseases

alpha-Mannosidosis

alpha-Mannosidosis

C16.320.565.595.577.500

C16.320.
C18.452.

C18.452.

beta-Mannosidosis

C16.320.565.595.577.750

C16.320.
C18.452.

C18.452.

Mucopolysaccharidoses

C16.320.565.595.600

C16.320.
C18.452.

C17.300.

Mucopolysaccharidosis I

C16.320.565.595.600.640

C16.320.
C18.452.

C17.300.

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.

Mucopolysaccharidosis IV

C16.320.565.595.600.655

C16.320.
C18.452.

C17.300.

Mucopolysaccharidosis VI

C16.320.565.595.600.670

C16.320.
C18.452.

C17.300.

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

Hemochromatosis

C16.320.565.618

C18.452.

Hepatolenticular Degeneration

C16.320.565.618.337

C18.452.

C18.452.

C16.320.565.618.403

C6.552.
C10.228.

C10.228.

C10.574.

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.

C13.351.

C18.452.

C18.452.

Familial Hypophosphatemic Rickets

C16.320.565.618.544.500

C5.116.
C13.351.

C12.777.

C18.452.

C16.320.

C18.452.

C18.452.

Menkes Kinky Hair Syndrome

C16.320.565.618.590

C10.228.
C10.574.

C10.574.

C10.597.

C16.320.

C16.320.

C16.320.

C17.800.

C18.452.

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.

C16.320.

C18.452.

Acatlasia

C16.320.565.663.25

C18.452.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

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. C10.314.
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
Mevalonate Kinase Deficiency**

C16.320.565.663.200
C16.320.565.663.480

C5.116. C18.452.
C10.228. C15.378.
C16.320. C16.320.
C18.452. C18.452.

Refsum Disease

C16.320.565.663.760

C18.452. C20.683.
C10.228. C10.500.
C10.574. C10.668.
C16.131. C16.320.
C16.320. C18.452.

Refsum Disease, Infantile

C16.320.565.663.865

C18.452. C18.452.
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, Variegata

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.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

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.
Familial Hypophosphatemic Rickets	C16.320.565.861.647.500	C5.116.	C12.777.
		C13.351.	C16.320.
		C18.452.	C18.452.
		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.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

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.
C16.614.
C17.800.
C17.800.
C17.800.
C18.452.

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

C16.320.565.925.500
C16.320.565.925.875

C18.452.
C16.131.
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.

Hamartoma Syndrome, Multiple

C16.320.700.435

C5.116. C5.116.
C4.445. C4.651.
C4.700.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

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. C4.700. C13.351. C19.344.	C4.588. C13.351. C17.800. C19.391.
Li-Fraumeni Syndrome	C16.320.700.600	C4.700.	C18.452.
Multiple Endocrine Neoplasia	C16.320.700.630	C4.588. C4.700.	C4.651. C19.344.
Multiple Endocrine Neoplasia Type 1	C16.320.700.630.500	C4.588.	C4.651.
Multiple Endocrine Neoplasia Type 2a	C16.320.700.630.505	C4.700.	C19.344.
Multiple Endocrine Neoplasia Type 2b	C16.320.700.630.510	C4.588. C4.700.	C4.651. C19.344.
Tuberous Sclerosis	C16.320.700.636	C4.445. C4.700.	C4.651. C10.500.
		C10.562. C16.131.	C10.574. C16.320.
Wilms Tumor	C16.320.700.642	C4.557. C4.700. C12.777.	C4.588. C12.758. C13.351.
		C13.351.	
Denys-Drash Syndrome	C16.320.700.642.220	C4.557. C4.700. C12.758.	C4.588. C12.706. C12.777.
		C13.351. C13.351. C16.131.	C13.351. C13.351. C16.131.
		C19.391.	
WAGR Syndrome	C16.320.700.642.950	C4.557. C4.700. C11.250. C11.941. C12.758.	C4.588. C10.597. C11.270. C12.706. C12.777.
		C13.351. C13.351. C16.131. C16.320. C19.391.	C13.351. C16.131. C16.131. C16.320. C16.320.
Neurofibromatoses	C16.320.700.645	C4.557. C10.562. C16.320.	C4.700. C10.574. C16.320.
Neurofibromatosis 1	C16.320.700.645.650	C4.557. C10.562.	C4.700. C10.574.
Neurofibromatosis 2	C16.320.700.645.655	C10.668. C4.557. C4.557. C9.218. C10.292. C10.562. C16.320.	C16.320. C4.557. C4.700. C9.647. C10.292. C10.574. C16.320.
Peutz-Jeghers Syndrome	C16.320.700.705	C4.700. C17.800.	C6.405.
Osteoarthropathy, Primary Hypertrophic	C16.320.718	C5.116.	C5.550.
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. C18.452.	C16.320.
Skin Diseases, Genetic	C16.320.850	C17.800.	

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Skin Diseases, Genetic

Albinism

Albinism	C16.320.850.80	C11.270. C16.320. C17.800.	C16.320. C17.800.
Albinism, Ocular	C16.320.850.80.90	C11.270. C16.320. C17.800.	C16.320. C17.800.
Albinism, Oculocutaneous	C16.320.850.80.100	C11.270. C16.320. C17.800.	C16.320. C17.800.
Hermanski-Pudlak Syndrome	C16.320.850.80.100.400	C11.270. C15.378. C15.378. C16.320. C17.800.	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.
Steatocystoma Multiplex	C16.320.850.250.856.500	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.
Epidermolysis Bullosa Simplex	C16.320.850.275.180	C16.131. C17.800.	C17.800. C17.800.
Erythrokeratoderma Variabilis	C16.320.850.337	C17.800.	C17.800.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Skin Diseases, Genetic

Hyalinosis, Systemic

Hyalinosis, Systemic	C16.320.850.368	C17.800.	
Ichthyosiform Erythroderma, Congenital	C16.320.850.400	C16.131. C17.800. C17.800.	C16.614. C17.800.
Hyperkeratosis, Epidermolytic	C16.320.850.400.375	C16.131. C17.800. 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.	C16.320. C17.800. C18.452.
Coproporphryia, Hereditary	C16.320.850.742.74	C6.552. C17.800. C18.452.	C16.320. C17.800. C18.452.
Porphyria, Acute Intermittent	C16.320.850.742.150	C6.552. C17.800. C18.452.	C16.320. C17.800. C18.452.
Porphyria Cutanea Tarda	C16.320.850.742.250	C6.552. C17.800. C18.452.	C16.320. C17.800. C18.452.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Genetic Diseases, Inborn

Skin Diseases, Genetic

Porphyrias, Hepatic

Porphyria, Hepatoerythropoietic

Porphyria, Hepatoerythropoietic

C16.320.850.742.437

C6.552.
C17.800.
C18.452.
C18.452.

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

Porphyria, Variegata

C16.320.850.742.625

C6.552.
C17.800.
C18.452.
C18.452.

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

Protoporphyrin, 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.
C18.452.

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.
C17.800.
C23.300.

Yellow Nail Syndrome

C16.320.962

C17.800.

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.

Congenital Hyperinsulinism

C16.614.200

C6.689.
C18.452.

C18.452.

Nesidioblastosis

C16.614.200.500

C6.689.
C18.452.

C18.452.

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.
C20.306.

Kernicterus

C16.614.304.502

C10.228.
C18.452.
C23.550.

C15.378.
C20.306.

Hernia, Umbilical

C16.614.378

C23.300.

Hydrophthalmos

C16.614.438

C11.250.
C16.131.

C11.525.

Hyperbilirubinemia, Neonatal

C16.614.451

C23.550.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities

Infant, Newborn, Diseases

Hyperbilirubinemia, Neonatal

Jaundice, Neonatal

Jaundice, Neonatal	C16.614.451.500	C23.550.	
Jaundice, Chronic Idiopathic	C16.614.451.500.250	C16.320.	C18.452.
Hyperostosis, Cortical, Congenital	C16.614.465	C5.116.	C5.116.
Ichthyosis	C16.614.492	C16.131.	C17.800.
		C17.800.	
Ichthyosiform Erythroderma, Congenital	C16.614.492.400	C16.131.	C16.320.
		C17.800.	C17.800.
		C17.800.	
Hyperkeratosis, Epidermolytic	C16.614.492.400.375	C16.131.	C16.320.
		C17.800.	C17.800.
		C17.800.	
Ichthyosis, Lamellar	C16.614.492.400.410	C16.131.	C16.320.
		C17.800.	C17.800.
		C17.800.	
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	C25.775.	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.	
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.

C16 - DISEASES-CONGENITAL, HEREDITARY, AND NEONATAL AND ABNORMALITIES

Congenital, Hereditary, and Neonatal Diseases and Abnormalities
Infant, Newborn, Diseases
Vitamin K Deficiency Bleeding

Vitamin K Deficiency Bleeding

C16.614.940

C15.378.

C15.378.

Wolman Disease

C16.614.947

C18.654.

C18.654.

C16.320.

C16.320.

C18.452.

C18.452.

C18.452.