

Am. Med. Soc.

EUROCAT GUIDE TO EPONYMS AND SYNDROMES

**FOR USE WITH
BPA Classification of Diseases
Paediatric Supplement to ICD 9**

**COMPILED BY
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PROJECT LEADER**

1979

FOREWORD

Many of the diseases of childhood, particularly those involving genetic and development abnormalities are classified in the International Classification of diseases (ICD 9th revision) to classes of somewhat but not always exactly similar conditions, and they cannot be certainly identified for retrieval purposes by using the ICD. The British Paediatric Association 5th digit extension of the ICD allows many individual syndromes to be identified, but in parts, still, different syndromes are classified together. The use of one extra digit (a 6th digit) allows the syndromes to be uniquely identified and the list in this book gives an appropriate numeric and alphabetic guide to these conditions, so far as the English version of the ICD goes.

However, it is anticipated that many terms in use in England, the USA and elsewhere have been omitted and the author will be grateful to be told of such terms and of their appropriate place within the International Classification. All languages are welcome, but particularly terms in use in European languages are of interest at present.

EUROCAT is an EEC programme for co-ordinating registers of congenital abnormalities in EEC countries and one aim of the study is to identify the diseases and terms which are truly synonymous.

I am very grateful for help given by Mr Adrian Read of the WHO Classification Unit in London who helped with the typing and checking and to Peggy Loy of the same unit who gave much valuable advice and to the staff of the Medical Statistics Unit of the Office of Population Censuses and Surveys, London who assisted with the checking.

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Project Leader of EUROCAT

EEC Concerted Action Project
EUROCAT
Registration of Congenital Abnormalities and Twins
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Sort in alphabetic order

1. Highlight Columns
A + B
2. Go to Data + sort
3. Column B Asc → Desc
4. Click OK.

LIST OF EPONYMS AND SYNDROMES IN BPA ADAPTATION OF ICD/9

015.0-(0)	Pott's disease
017.6-(0)	Addison's disease Tuberculosis
017.6-(0)	Tuberculosis Addison's disease
036.3-(0)	Waterhouse Friderichsen's syndrome
040.2-(0)	Whipple's disease
046.1-(0)	Jakob-Creutzfeldt disease
053.1-(0)	Ramsey-Hunt syndrome
054.0-(0)	Kaposi's disease, meaning varicelliform eruption
054.0-(0)	Kaposi's varicelliform eruption
074.1-(0)	Bornholm's disease
078.8-(0)	Marburg fever
081.1-(0)	Brill-Zinsser disease
086.2-(0)	Chagas's disease
090.5-(0)	Hutchinson's teeth
099.2-(0)	Donovanosis
099.3-(0)	Reiter's syndrome
099.3-(1)	Reiter's conjunctivitis
100.0-(0)	Weill's disease
101.--(0)	Vincent's angina
101.--(0)	Vincent's stomatitis
125.2-(0)	Calabar swelling
136.1-(0)	Behcet's syndrome
157.4-(0)	Langerhans, islets of, neoplasm, malignant
173.9-(0)	Kaposi's sarcoma
189.0-(0)	Wilm's tumour
200.2-(0)	Burkitt's lymphoma or tumour
201.0-(0)	Hodgkin's paragranuloma

201.1-(0) Hodgkin's granuloma
 201.2-(0) Hodgkin's sarcoma
 201.4-(0) Hodgkin's histiocytic predominance granuloma
 201.5-(0) Hodgkin's nodular sclerosis
 201.6-(0) Hodgkin's mixed cellularity disease
 201.7-(0) Hodgkin's depletion granuloma
 201.9-(0) Hodgkin's disease
 201.9-(0) Hodgkin's lymphoma
 202.2-(0) Sezary's disease
 202.5-(0) Letterer-Siwe disease
 207.0-(0) Di Guglielmo's disease
 211.7-(0) Langerhans, islets of, neoplasm, benign
 216.9-(0) Jadassohn-Tieche naevus
 216.9-(1) Blue naevus
 216.9-(2) Hairy naevus
 216.9-(3) Junctional naevus
 230.9-(0) Langerhans, islets of, neoplasm, in situ
 235.5-(0) Langerhans, islets of, neoplasm, uncertain behaviour
 237.7-(0) Von Recklinghausen's disease
 239.0-(0) Langerhans, islets of, neoplasm, unspecified nature
 243.90(0) Pendred's syndrome
 245.2-(0) Hashimoto's disease
 245.2-(1) Hashimoto's thyroiditis
 251.5-(0) Zollinger-Ellison syndrome
 253.80(0) Rathke's pouch, cyst
 253.81(0) Frohlich's syndrome
 255.0-(0) Cushing's disease
 255.0-(1) A.C.T.H. ectopic syndrome
 255.10(0) Conn's syndrome

255.25(0) Adrenogenital (congenital) syndrome with salt loss
 255.26(0) Adrenogenital (congenital) syndrome
 255.40(0) Addison's disease
 256.4-(0) Stein-Leventhal syndrome
 257.21(0) Kallman's syndrome
 259.80(0) Progeria
 259.82(0) Leprechaunism
 259.83(0) Werner's disease or syndrome
 270.02(0) Fanconi(-de Toni) (-Debre) syndrome
 270.02(0) De Toni-Fanconi (-Debre) syndrome
 270.03(0) Hartnup's syndrome
 270.20(0) Klein-Waardenburg syndrome
 270.29(0) Lowe's syndrome
 270.31(0) Maple syrup (urine) disease
 271.00(0) Von Gierke's disease
 271.01(0) Pompe's disease
 271.04(0) McArdle's syndrome
 272.5-(0) Bassen-Kornzweig disease
 272.5-(0) Tangier disease
 272.7-(0) Fabry's disease
 272.7-(1) Gaucher's disease
 272.7-(2) I-cell disease
 272.7-(3) Niemann-Pick disease
 272.7-(4) Wolman's disease
 275.02(0) Von Recklinghausen-Applebaum disease
 275.1-(0) Wilson's disease
 277.2-(0) Lesch-Nyhan disease or syndrome
 277.40(0) Crigler-Najjar syndrome

277.42(0) Gilbert's syndrome
 277.43(0) Dubin-Johnson syndrome
 277.43(0) Rotor's syndrome
 277.51(0) Hurler's syndrome
 277.51(1) Scheie's syndrome
 277.52(0) Hunter's syndrome
 277.53(0) Sanfilippo's syndrome
 277.54(0) Morquio's disease
 277.56(0) Maroteaux-Lamy syndrome
 277.87(0) Hand-Schuller-Christian syndrome
 279.10(0) Di George syndrome
 279.11(0) Wiskott-Aldrich disease or syndrome
 282.49(0) Cooley's anaemia
 284.00(0) Fanconi's (aplastic) anaemia
 284.02(0) Blackfan-Diamond anaemia or syndrome
 286.1-(0) Christmas disease
 286.4-(0) Von Willebrand's disease
 287.0-(0) Henoch(-Schonlein) purpura
 287.1-(0) Glanzmann's syndrome
 287.2-(0) Gardner-Diamond syndrome
 288.2-(0) Chediak-Higashi syndrome
 299.0-(0) Kanner's syndrome
 299.1-(0) Heller's syndrome
 300.1-(0) Ganer's (hysterical) syndrome
 307.2-(0) Gilles de la Tourette syndrome
 330.0-(0) Canavan's disease
 330.0-(1) Pelizaeus-Merzbacher disease
 330.0-(2) Krabbe's disease or leukodystrophy
 330.1-(0) Jansky-Bielchowsky disease

330.1-(1) Kuf's disease
 330.1-(2) Spellmeyer-Vogt disease
 330.1-(3) Tay-Sach's disease
 330.1-(4) Batten's disease
 330.8-(0) Alper's disease
 330.8-(1) Leigh's disease, encephalomyelopathy or syndrome
 331.8-(0) Reye's disease
 333.22(0) Unverricht-Lundborg disease
 333.4-(0) Huntington's chorea
 333.70(0) Vogt's disease
 334.0-(0) Friedreich's ataxia
 334.3-(0) Marinesco-Sjogren syndrome
 334.3-(1) Roussy-Levy syndrome
 334.8-(0) Louis-Bar syndrome
 335.0-(0) Werdnig-Hoffman disease
 335.1-(0) Kugelberg-Welander syndrome
 337.9-(0) Horner's syndrome
 341.0-(0) Devic's disease
 341.1-(0) Schilder's disease
 341.1-(1) Baló's concentric sclerosis
 344.80(0) Todd's paralysis
 345.50(0) Jacksonian epilepsy
 346.8-(0) Moebius disease
 351.0-(0) Bell's palsy
 352.9-(0) Bertolotti's syndrome
 356.0-(0) Dejerine-Sottas neuropathy*
 356.1-(0) Charcot-Marie-Tooth disease
 356.3-(0) Refsum's disease or syndrome

357.0-(0) Guillain-Barre syndrome
 358.8-(0) Floppy infant syndrome
 358.8-(0) Oppenheim's disease
 359.10(0) Duchenne (type) hereditary muscular dystonia
 359.11(0) Becker's disease
 362.72(0) Leber's amaurosis
 371.1-(0) Kayser-Fleischer ring
 371.3-(0) Descemet's membrane, fold or rupture
 378.7-(0) Duane's syndrome
 378.7-(1) Stilling's syndrome
 378.7-(2) Turck's syndrome
 379.4-(0) Adie(-Holmes) syndrome
 386.0-(0) Meniere's syndrome
 392.9-(0) Sydenham's chorea
 424.9-(0) Libman-Sacks endocarditis
 426.1-(0) Wenkebach's phenomenon
 426.5-(0) His, bundle of, interruption of
 426.7-(0) Wolff-Parkinson-White syndrome
 426.8-(0) Jervell-Lange-Neilson syndrome
 426.8-(1) Lange-Neilson syndrome
 426.8-(2) Lown-Ganong-Levine syndrome
 437.5-(0) Moyamoya syndrome
 443.0-(0) Raynaud's syndrome
 446.2-(0) Goodpasture's syndrome
 446.4-(0) Wegener's granulomatosis
 446.7-(0) Takayasu's arteritis, disease or syndrome
 448.0-(0) Osler-Rendu-Weber syndrome
 448.0-(1) Rendu-Osler-Weber syndrome
 453.0-(0) Budd-Chiari syndrome

453.0-(1) Stuart-Brass syndrome
 516.3-(0) Hamman-Rich syndrome
 518.3-(0) Loeffler's syndrome
 527.1-(0) Mikulicz's disease
 530.7-(0) Mallory-Weiss syndrome
 555.9-(0) Crohn's disease
 571.2-(0) Laennec's Cirrhosis-Alcoholic
 571.59(0) Laennec's Cirrhosis non-Alcoholic
 579.2-(0) Blind loop syndrome
 583.26(0) Berger's disease
 588.80(0) Lightwood-Albright syndrome
 593.89(0) Bartter's syndrome
 691.80(0) Besnier's eczema or prurigo
 694.4-(0) Usher - Senear disease
 695.11(0) Lyell's syndrome
 695.12(0) Stevens-Johnson syndrome
 695.81(0) Ritter's disease
 695.82(0) Leiner's disease
 705.8-(0) Fox(-Fordyce) disease
 710.2-(0) Sjogren's disease
 714.1-(0) Felty's syndrome
 714.3-(0) Still's disease
 714.4-(0) Jaccoud's syndrome
 732.0-(0) Scheuermann's disease
 732.1-(0) Legg-Calve-Perthes disease
 732.4-(0) Blount's disease
 732.4-(1) Kohler's disease (patellar)
 732.4-(2) Osgood-Schlatter disease

732.5-(0) Kohler's disease (tarsal navicular)
 733.6-(0) Tietze's disease
 741.01(0) Arnold-Chiari syndrome
 741.08(0) Dandy Walker syndrome with spina bifida
 741.98(0) Billroth's disease
 742.30(0) Aqueduct of Sylvius' anomaly
 742.30(0) Sylvius' aqueduct anomaly
 742.31(1) Dandy-Walker syndrome
 742.31(0) Magendie and Luschka anomaly
 742.80(1) Jaw winking phenomenon
 742.80(0) Marcus-Gunn's syndrome
 742.80(0) Gunn's syndrome
 742.81(1) Familial dysautonomia
 742.81(0) Riley-Day syndrome
 743.32(0) Blue dot cataract
 743.45(0) Blue sclera
 743.48(0) Reiger's anomaly
 744.03(0) Corti organ, anomaly of
 744.23(0) Vulcan ear(s)
 744.25(0) Eustachian tube, anomaly of
 744.28(0) Darwin's tubercle
 745.11(0) Taussig-Bing complex
 745.20(0) Fallot's tetrad or tetralogy
 745.21(0) Fallot's pentalogy
 745.40(0) Roger's disease
 745.41(0) Eisenmenger's defect
 745.42(0) Gerbode defect
 745.50(0) Foramen ovale, patent non-closure
 745.50(0) Foramen ovale

745.51(0) Ostium secundum
 745.52(0) Lutembacher's syndrome
 745.60(0) Ostium primum
 746.2-(0) Ebstein's anomaly
 746.5-(0) Duroziez's disease
 746.84(0) Fallot's trilogy
 746.86(0) Uhl's disease
 746.88(0) Eustachian valve, non-closure
 746.92(0) Acyanotic heart disease
 746.93(1) Blue baby syndrome
 746.93(2) Cyanotic heart disease or syndrome
 746.93(0) Blue syndrome
 747.0-(0) Ductus botalli anomaly
 747.0-(0) Botalli ductus anomaly
 747.24(0) Valsalva, sinus of, anomalies
 750.22(0) Ranula
 750.23(0) Wharton's duct, anomaly or displacement of
 750.28(0) Fordyce's disease (mouth)
 751.01(0) Meckel's diverticulum anomaly
 751.33(0) Hirschsprung's disease
 751.41(0) Jackson's membrane or veil
 751.58(0) Short bowel syndrome
 752.11(0) Gartner's duct anomaly
 752.47(0) Nuck, canal of, anomaly of
 752.87(0) Morgagni, hydatid of
 752.87(0) Hydatid of Morgagni
 752.87(1) Wolffian cyst
 753.00(0) Potter's syndrome
 753.13(0) Polycystic renal disease or syndrome

754.01(0) Potter's facies
 754.88(0) Guerin-Stern syndrome
 755.50(1) Acrocephalosyndactyly
 755.50(0) Apert's syndrome
 755.50(2) Macroductylia
 755.51(0) Lobster claw hand
 755.52(0) Madelung's deformity
 755.55(1) Cleidocranial dysostosis
 755.55(0) Sprengel's deformity
 755.61(1) Lobster Claw foot
 755.61(0) Rocker bottom feet
 755.81(0) Larsen's syndrome
 756.01(0) Crouzon's disease
 756.03(0) Pierre Robin syndrome
 756.03(0) Robin's syndrome
 756.04(2) Mandibulo-facial dysostosis
 756.04(0) Franceschetti's syndrome
 756.04(1) Treacher-Collins syndrome
 756.05(1) Oculomandibular dysostosis
 756.05(0) Hallerman-Streif syndrome
 756.05(2) Eyelid malar mandible syndrome
 756.06(0) Goldenhaar's syndrome
 756.08(0) First arch syndrome
 756.11(0) Feil-Klippel syndrome
 756.11(0) Klippel-Feil syndrome
 756.11(1) Wildervanck's syndrome
 756.18(1) Straight back syndrome
 756.40(0) Jeune's syndrome
 756.41(0) Ollier's disease

756.42(0) Kast's syndrome
 756.42(1) Maffucci syndrome
 756.44(1) Diastrophic dwarf
 756.44(2) Metatrophic dwarf
 756.44(3) Thanatotropic dwarf
 756.44(0) Emery-Nelson syndrome
 756.50(0) Adair-Dighton syndrome
 756.50(4) Van der Hoeve's syndrome
 756.50(5) Vrolik's disease
 756.50(2) Eddowes's syndrome
 756.50(0) Dighton-Adair syndrome
 756.51(0) Albright(-McCune)-Sternberg syndrome
 756.51(2) McCune-Albright(-Sternberg) syndrome
 756.51(1) Sternberg-McCune syndrome
 756.50(1) Lobstein disease
 756.52(0) Ellis-van Creveld syndrome
 756.53(0) Caffey's syndrome
 756.54(0) Albers-Schonberg syndrome
 756.54(1) Marble bone syndrome
 756.55(1) Camurati-Englemann disease
 756.55(0) Blue sclera syndrome (Blue sclera with fragility of bones and deafness)
 756.55(2) Englemann's disease
 756.57(0) Conradi-Huberman syndrome
 756.57(1) Conradi's syndrome
 756.58(1) Leri-Weill syndrome
 756.58(0) Furst-Ostrum syndrome
 756.58(0) Ostrum-Furst syndrome
 756.72(0) Prune belly syndrome

756.80(0) Poland's syndrome
 756.81(0) Abdominal muscle syndrome
 756.81(1) Orbinsky's syndrome
 756.83(0) Nail patella syndrome
 756.85(1) Danlos' syndrome
 756.85(0) Ehlers-Danlos syndrome
 756.88(4) Ayala's disease
 756.88(0) Schwartz's syndrome
 756.88(1) Catel-Schwartz-Jampel syndrome
 756.88(2) Osterreicher-Turner syndrome
 756.88(3) Soto's syndrome
 756.88(5) Popliteal web syndrome
 757.0-(0) Milroy's disease
 757.0-(1) Meige's disease
 757.10(0) Harlequin fetus
 757.11(0) Collodion baby
 757.12(0) Sjogren-Larsson syndrome
 757.30(6) Darier's disease
 757.30(0) Hailey-Hailey disease
 757.30(1) Brusch's acromicria, actopachycardia, acropachyderma or dystrophia-osteogenitalis
 757.30(2) Thomson's disease or syndrome
 757.30(3) Rothmund's syndrome
 757.30(3) Rothmund-Thomson disease or syndrome
 757.30(4) Sieman's syndrome
 757.30(5) Mibelli angiokeratoma or hyperkeratosis
 757.30(7) Telangiectasic pigmentation cataract
 757.32(0) Nettleship's syndrome
 757.33(2) Goldscheider's disease

757.33(1) Fox's disease
 757.33(4) Koebner's disease
 757.35(0) Bloch-Sulzberger syndrome
 757.36(0) Kaposi's disease
 757.38(0) Mongolia blue spot
 757.39(1) Mast cell disease
 757.39(2) Mastocytosis
 757.43(0) Taenzer's disease
 757.8-(0) Bloom(-Machacek) (-Torr) syndrome
 758.00(0) Trisomy 21 syndrome
 758.01(0) Trisomy G syndrome
 758.09(1) Langdon-Down syndrome
 758.09(0) Down's syndrome
 758.10(0) Trisomy 13 syndrome
 758.11(0) Trisomy D Nos syndrome
 758.19(0) Patau's syndrome
 758.20(0) Trisomy 18 syndrome
 758.21(0) Trisomy E Nos syndrome
 758.29(0) Edwards's syndrome
 758.30(0) Antimongolism syndrome
 758.31(0) Cat cry syndrome
 758.31(0) Cri du chat syndrome
 758.31(1) Cri du chat (meaning deletion of Karyotype 5 short arm 5 or B)
 758.32(0) Wolff-Hirschorn syndrome
 758.39(0) Autosomal deletion syndrome
 758.52(0) Trisomy syndrome Nos
 758.60(2) Turner's karyotype 45
 758.60(1) Turner's syndrome (XO) Karyotype
 758.61(0) XO mosaic (Turner's phenotype) syndrome

758.61(1) Partial X deletion syndrome
 758.61(2) Ring chromosome
 758.61(3) Turner's phenotype, isochromosome
 758.61(4) Turner's phenotype, variant karyotype
 758.61(5) Turner's phenotype, XO/XX mosaic
 758.69(0) Bonnevie-Ullrich syndrome
 758.69(1) Turner's syndrome
 758.69(2) Pseudo-Turner syndrome
 758.69(0) Ullrich(-Bonnevie)-Turner syndrome
 758.70(0) XXY syndrome
 758.71(1) Klinefelter's XXYY syndrome
 758.71(0) Klinefelter's XXXXY syndrome
 758.79(0) Klinefelter's syndrome
 758.85(0) Triple X female syndrome
 758.85(1) XXX female osteodystrophy
 758.88(0) Sex chromosome mosaic
 758.88(0) Mosaic, sex
 758.88(1) Penta X
 758.90(0) Mosaic Nos
 759.34(0) Kartagener's syndrome
 759.34(1) Triad disease or syndrome
 759.5-(1) Pringle's disease or syndrome
 759.5-(0) Bourneville's disease
 759.60(0) Peutz-Jeghers syndrome
 759.61(0) Kalischer syndrome
 759.61(1) Sturge-Weber(-Dimitri) syndrome
 759.61(2) Sturge-Kalischer-Weber syndrome
 759.62(0) Von Hippel (-Lindau) syndrome
 759.63(0) Gardner's syndrome

759.80(5) Meyer-Schwickerath - Weyers syndrome
 759.80(7) Freeman-Sheldon syndrome
 759.80(3) Whistling face syndrome
 759.80(0) Mohr's syndrome
 759.80(1) Cyclops syndrome
 759.80(2) Oro-facial-digital syndrome
 759.80(4) Waardenburg's syndrome
 759.82(6) Cockayne's syndrome
 759.82(7) Seckel's syndrome
 759.82(3) Silver's syndrome
 759.82(3) Russell-Silver syndrome
 759.82(2) Laurence-Moon-Biedl syndrome
 759.82(1) Cornelia de Lange syndrome
 759.82(1) Amsterdam dwarfism
 759.82(1) De Lange's syndrome
 759.82(0) Biedle-Bardet syndrome
 759.82(8) Smith-Lemli-Opitz syndrome
 759.82(5) Bruck-de Lange syndrome
 759.84(0) Klippel-Trenaunay(-Weber) syndrome
 759.84(1) Rubinstein-Taybi syndrome
 759.84(2) Holt-Oram syndrome
 759.84(3) Carpenter's syndrome
 759.84(4) Sirenomelia
 759.84(6) Aglossa-adactyly
 759.84(5) Otopalatodigital syndrome
 759.86(0) Marfan's syndrome
 759.86(1) Arachnodactyly syndrome
 759.87(0) Alport's syndrome
 759.87(1) Biemond syndrome

759.87(2) Prader-Willi syndrome
 759.87(3) Menke's (kinky hair) syndrome
 759.87(4) Beckwith(-Wiedemann) syndrome
 759.87(5) Zellweger's disease or syndrome
 759.87(6) Oculo-cerebral-renal syndrome
 759.89(0) Ullrich-Feichtiger syndrome
 759.89(1) Polycystic syndrome
 759.89(1) Polycystic disease (liver and kidney)
 759.89(2) Fraser's syndrome
 759.89(3) Papillon-Leage and Psaume syndrome
 759.89(4) Usher's syndrome
 759.89(5) VATER syndrome
 759.89(6) Noonan's syndrome
 759.89(7) Marchesani(-Weill) syndrome
 759.89(8) Gorlin-Chaudry-Moss syndrome
~~759.89(9) Happy puppet~~
 767.60(0) Erb's palsy
 767.60(1) Erb(-Duchenne) paralysis
 767.61(0) Klumpke's paralysis or palsy
 770.70(0) Wilson-Mikity syndrome
 771.0-(0) Rubella syndrome
 774.31(0) Lucey-Driscoll syndrome
 775.0-(0) Infant of diabetic mother syndrome
 778.2-(0) Cold injury, newborn syndrome
 778.82(0) Bronze baby syndrome
 779.5-(0) Drug withdrawal (newborn) syndrome
 798.0-(0) Sudden infant death syndrome
 995.1-(0) Quinke's oedema
 995.50(0) Battered baby syndrome

ALPHABETICAL LISTING OF EPONYMS, SYNDROMES, etc.

Abdominal muscle syndrome 756.81(0)
 Acrocephalosyndactyly 755.50(1)
 A C T H ectopic syndrome 255.0-(1)
 Acyanotic heart disease 746.92(0)
 Adair-Dighton syndrome 756.50(0)
 Addison's disease 255.40(0)
 Addison's disease, tuberculous 017.6-(0)
 Adiel(-Holmes) syndrome 379.4-(0)
 Adrenogenital (congenital) syndrome 255.26(0)
 Adrenogenital (congenital) syndrome with salt loss 255.25(0)
 Aglosso-adactyly 759.84(6)
 Albers-Schonberg syndrome 756.54(0)
 Albright(-McCune)-Sternberg syndrome 756.51(0)
 Alper's disease 330.8-(0)
 Alport's syndrome 759.87(0)
 Amsterdam dwarfism 759.82(1)
 Antimongolism syndrome 758.30(0)
 Apert's syndrome 755.50(0)
 Aqueduct of Sylvius' anomaly 742.30(0)
 Arachnodactyly syndrome 759.86(1)
 Arnold-Chiari syndrome 741.01(0)
 Autosomal deletion syndrome 758.39(0)
 Ayala's disease 756.88(4)
 Balo's concentric sclerosis 341.1-(1)
 Bartter's syndrome 593.89(0)
 Bassen-Kornzweig disease 272.5-(0)
 Batten's disease 330.1-(4)
 Battered baby syndrome 995.50(0)

Becker's disease 359.11(0)
 Beckwith(-Wiedemann) syndrome 759.87(4)
 Behcet's syndrome 136.1-(0)
 Bell's palsy 351.0-(0)
 Berger's disease 583.26(0)
 Bertolotti's syndrome 352.9-(0)
 Besnier's eczema or prurigo 691.80(0)
 Biedle-Bardet syndrome 759.82(0)
 Biemond syndrome 759.87(1)
 Billroth's disease 741.98(0)
 Blackfan-Diamond anaemia or syndrome 284.02(0)
 Blind loop syndrome 579.2-(0)
 Bloch-Sulzberger syndrome 757.35(0)
 Bloom(-Machacek)(-Torr) syndrome 757.8-(0)
 Blount's disease 732.4-(0)
 Blue baby syndrome 746.93(1)
 Blue dot cataract 743.32(0)
 Blue naevus 216.9-(1)
 Blue sclera 743.45(0)
 Blue sclera syndrome [Blue sclera with fragility of bones and deafness] 756.55(0)
 Blue syndrome 746.93(0)
 Bonnevie-Ullrich syndrome 758.69(0)
 Bornholm's disease 074.1-(0)
 Botalli ductus anomaly 747.0-(0)
 Bourneville's disease 759.5-(0)
 Brill-Zinsser disease 081.1-(0)
 Bronze baby syndrome 778.82(0)
 Bruck-de Lange syndrome 759.82(5)
 Brusch's acromicria, actopachycardia, acropachyderma or dystrophia-osteogenitalis 757.30(1)
 Budd-Chiari syndrome 453.0-(0)

Burkitt's lymphoma or tumour 200.2-(0)
 Caffey's syndrome 756.53(0)
 Calabar swelling 125.2-(0)
 Camurati-Englemann disease 756.55(1)
 Canavan's disease 330.0-(0)
 Carpenter's syndrome 759.84(3)
 Cat cry syndrome 758.31(0)
 Catel-Schwartz-Jampel syndrome 756.88(1)
 Chagas's disease 086.2-(0)
 Charcot-Marie-Tooth disease 356.1-(0)
 Chediak-Higashi syndrome 288.2-(0)
 Christmas disease 286.1-(0)
 Cleidocranial dysostosis 755.55(1)
 Cockayne's syndrome 759.82(6)
 Cold injury, newborn syndrome 778.2-(0)
 Collodion baby 757.11(0)
 Conn's syndrome 255.10(0)
 Conradi's syndrome 756.57(1)
 Conradi-Huberman syndrome 756.57(0)
 Cooley's anaemia 282.49(0)
 Cornelia de Lange syndrome 759.82(1)
 Corti organ, anomaly of 744.03(0)
 Cri du chat syndrome 758.31(0)
 Cri du chat (meaning deletion of karyotype 5, short arm 5 or B) 758.31(1)
 Crigler-Najjar syndrome 277.40(0)
 Crohn's disease 555.9-(0)
 Crouzon's disease 756.01(0)
 Cushing's disease 255.0-(0)
 Cyanotic heart disease or syndrome 746.93(2)

Cyclops syndrome 759.80(1)

Dandy Walker syndrome 742.31(1)

Dandy Walker syndrome with spina bifida 741.08(0)

Danlos' syndrome 756.85(1)

Darier's disease 757.30(6)

Darwin's tubercle 744.28(0)

Dejerine-Sottas neuropathy 356.0-(0)

De Lange's syndrome 759.82(1)

Descemet's membrane, fold or rupture 371.3-(0)

De Toni-Fanconi(-Debre) syndrome 270.02(0)

Devic's disease 341.0-(0)

Diastrophic dwarf 756.44(1)

Di George syndrome 279.10(0)

Dighton-Adair syndrome 756.50(0)

Di Guglielmo's disease 207.0-(0)

Donovanosis 099.2-(0)

Down's syndrome 758.09(0)

Drug withdrawal (newborn) syndrome 779.5-(0)

Duane's syndrome 378.7-(0)

Dubin-Johnson syndrome 277.43(0)

Duchenne (type) hereditary muscular dystonia 359.10(0)

Ductus botalli anomaly 747.0-(0)

Duroziez's disease 746.5-(0)

Ebstein's anomaly 746.2-(0)

Eddowes's syndrome 756.50(2)

Edwards's syndrome 758.29(0)

Ehlers-Danlos syndrome 756.85(0)

Eisenmenger's defect 745.41(0)

Ellis-van Creveld syndrome 756.52(0)

Emery-Nelson syndrome 756.44(0)

Engelmann's disease 756.55(2)

Erb(-Duchenne) paralysis 767.60(1)

Erb's palsy 767.60(0)

Eustachian tube, anomaly of 744.25(0)

Eustachian valve, nonclosure 746.88(0)

Eyelid malar mandible syndrome 756.05(2)

Fabry's disease 272.7-(0)

Fallot's pentalogy 745.21(0)

Fallot's tetrad or tetralogy 745.20(0)

Fallot's trilogy 746.84(0)

Familial dysautonomia 742.81(1)

Fanconi's (aplastic) anaemia 284.00(0)

Fanconi(-de Toni)(-Debre) syndrome 270.02(0)

Feil-Klippel syndrome 756.11(0)

Felty's syndrome 714.1-(0)

First arch syndrome 756.08(0)

Floppy infant syndrome 358.8-(0)

Foramen ovale 745.50(0)

Foramen ovale, patent nonclosure 745.50(0)

Fordyce's disease (mouth) 750.28(0)

Fox(-Fordyce) disease 705.8-(0)

Fox's disease 757.33(1)

Franceschetti's syndrome 756.04(0)

Fraser's syndrome 759.89(2)

Freeman-Sheldon syndrome 759.80(7)

Friedreich's ataxia 334.0-(0)

Frohlich's syndrome 253.81(0)

Furst-Ostrum syndrome 756.58(0)

Ganster's (hysterical) syndrome 300.1-(0)
 Gardner-Diamond syndrome 287.2-(0)
 Gardner's syndrome 759.63(0)
 Gartner's duct anomaly 752.11(0)
 Gaucher's disease 272.7-(1)
 Gerbode defect 745.42(0)
 Gilbert's syndrome 277.42(0)
 Gilles de la Tourette syndrome 307.2-(0)
 Glanzmann's syndrome 287.1-(0)
 Goldenhaar's syndrome 756.06(0)
 Goldscheider's disease 757.33(2)
 Goodpasture's syndrome 446.2-(0)
 Gorlin-Chaudry-Moss syndrome 759.89(8)
 Guerin-Stern syndrome 754.88(0)
 Guillain-Barre syndrome 357.0-(0)
 Gunn's syndrome 742.80(0)

 Hailey-Hailey disease 757.30(0)
 Hairy naevus 216.9-(2)
 Hallerman-Streif syndrome 756.05(0)
 Hamman-Rich syndrome 516.3-(0)
 Hand-Schuller-Christian syndrome 277.87(0)
 Happy puppet 759.89(9)
 Harlequin fetus 757.10(0)
 Hartnup's syndrome 270.03(0)
 Hashimoto's disease 245.2-(0)
 Hashimoto's thyroiditis 245.2-(1)
 Heller's syndrome 299.1-(0)
 Henoch(-Schonlein) purpura 287.0-(0)
 Hirschsprung's disease 751.33(0)

His, bundle of, interruption of 426.5-(0)
 Hodgkin's depletion granuloma 201.7-(0)
 Hodgkin's disease 201.9-(0)
 Hodgkin's granuloma 201.1-(0)
 Hodgkin's histiocytic predominance granuloma 201.4-(0)
 Hodgkin's lymphoma 201.9-(0)
 Hodgkin's mixed cellularity disease 201.6-(0)
 Hodgkin's nodular sclerosis 201.5-(0)
 Hodgkin's paraganuloma 201.0-(0)
 Hodgkin's sarcoma 201.2-(0)
 Holt-Oram syndrome 759.84(2)
 Horner's syndrome 337.9-(0)
 Hunter's syndrome 277.52(0)
 Huntington's chorea 333.4-(0)
 Hurler's syndrome 277.51(0)
 Hutchinson's teeth 090.5-(0)
 Hydatid of morgagni 752.87(0)

 I-cell disease 272.7-(2)
 Infant of diabetic mother syndrome 775.0-(0)

 Jaccoud's syndrome 714.4-(0)
 Jacksonian epilepsy 345.50(0)
 Jackson's membrane or veil 751.41(0)
 Jadassohn-Tieche naevus 216.9-(0)
 Jakob-Creutzfeldt disease 046.1-(0)
 Jansky-Bielchowsky disease 330.1-(0)
 Jaw winking phenomenon 742.80(1)
 Jervell-Lange-Neilson syndrome 426.8-(0)
 Jeune's syndrome 756.40(0)

Junctional naevus 216.9-(3)

Kalischer syndrome 759.61(0)

Kallman's syndrome 257.21(0)

Kanner's syndrome 299.0-(0)

Kaposi's disease 757.36(0)

Kaposi's disease, meaning varicelliform eruption 054.0-(0)

Kaposi's sarcoma 173.9-(0)

Kaposi's varicelliform eruption 054.0-(0)

Kartagener's syndrome 759.34(0)

Kast's syndrome 756.42(0)

Kayser-Fleischer ring 371.1-(0)

Klein-Waardenburg syndrome 270.20(0)

Klinefelter's syndrome 758.79(0)

Klinefelter's XXXXY syndrome 758.71(0)

Klinefelter's XYY syndrome 758.71(1)

Klippel-Feil syndrome 756.11(0)

Klippel-Trenaunay(-Weber) syndrome 759.84(0)

Klumpke's paralysis or palsy 767.61(0)

Koebner's disease 757.33(4)

Kohler's disease (patellar) 732.4-(1)

Kohler's disease (tarsal navicular) 732.5-(0)

Krabbe's disease or leukodystrophy 330.0-(2)

Kuf's disease 330.1-(1)

Kugelberg-Welander syndrome 335.1-(0)

Laennec's cirrhosis, alcoholic 571.2-(0)

Laennec's cirrhosis, non-alcoholic 571.2-(0)

Langdon-Down syndrome 758.09(1)

Lange-Neilson syndrome 426.8-(1)

Langerhans, islets of, neoplasm, benign 211.7-(0)

Langerhans, islets of, neoplasm, in situ 230.9-(0)

Langerhans, islets of, neoplasm, malignant 157.4-(0)

Langerhans, islets of, neoplasm, uncertain behaviour 235.5-(0)

Langerhans, islets of, neoplasm, unspecified nature 239.0-(0)

Larsen's syndrome 755.81(0)

Laurence-Moon-Biedl syndrome 759.82(2)

Leber's amaurosis 362.72(0)

Legg-Calve-Perthes disease 732.1-(0)

Leigh's disease, encephalomyelopathy or syndrome 330.8-(1)

Leiner's disease 695.82(0)

Leprechaunism 259.82(0)

Leri-Weill syndrome 756.58(1)

Lesch-Nyhan disease or syndrome 277.2-(0)

Letterer-Siwe disease 202.5-(0)

Libman-Sacks endocarditis 424.9-(0)

Lightwood-Albright syndrome 588.80(0)

Lobstein disease 756.50(1)

Lobster claw foot 755.61(1)

Lobster claw hand 755.51(0)

Loeffler's syndrome 518.3-(0)

Louis-Bar syndrome 334.8-(0)

Lowe's syndrome 270.29(0)

Lown-Ganong-Levine syndrome 426.8-(2)

Lucey-Driscoll syndrome 774.31(0)

Lutembacher's syndrome 745.52(0)

Lyell's syndrome 695.11(0)

Macroductylia 755.50(2)

Madelung's deformity 755.52(0)

Maffucci syndrome 756.42(1)
 Magendie and Luschka anomaly 742.31(0)
 Mallory-Weiss syndrome 530.7-(0)
 Mandibulo-facial dysostosis 756.04(2)
 Maple syrup (urine) disease 270.31(0)
 Marble bone syndrome 756.54(1)
 Marburg fever 078.8-(0)
 Marchesani(-Weill) syndrome 759.89(7)
 Marcus-Gunn syndrome 742.80(0)
 Marfan's syndrome 759.86(0)
 Marinesco-Sjogren syndrome 334.3-(0)
 Maroteaux-Lamy syndrome 277.56(0)
 Mast cell disease 757.39(1)
 Mastocytosis 757.39(2)
 McArdle's syndrome 271.04(0)
 McCune-Albright(-Sternberg) syndrome 756.51(2)
 Meckel's diverticulum anomaly 751.01(0)
 Meige's disease 757.0-(1)
 Meniere's syndrome 386.0-(0)
 Menke's (kinky hair) syndrome 759.87(3)
 Metatrophic dwarf 756.44(2)
 Meyer-Schwickerath-Weyers syndrome 759.80(5)
 Mibelli angiokeratoma or hyperkeratosis 757.30(5)
 Mikulicz's disease 527.1-(0)
 Milroy's disease 757.0-(0)
 Moebius disease 346.8-(0)
 Mohr's syndrome 759.80(0)
 Mongolian blue spot 757.38(0)
 Morgagni, hydatid of 752.87(0)
 Morquio's disease 277.54(0)

Mosaic NOS 758.90(0)
 Mosaic, sex 758.88(0)
 Moyamoya syndrome 437.5-(0)

 Nail patella syndrome 756.83(0)
 Nettleship's syndrome 757.32(0)
 Niemann-Pick disease 272.7-(3)
 Noonan's syndrome 759.89(6)
 Nuck, Canal of, anomaly of 752.47(0)

 Oculo-cerebral-renal syndrome 759.87(6)
 Oculomandibular dysostosis 756.05(1)
 Ollier's disease 756.41(0)
 Oppenheim's disease 356.8-(0)
 Orbinsky's syndrome 756.81(1)
 Oro-facial-digital syndrome 759.80(2)
 Osgood-Schlatter disease 732.4-(2)
 Osler-Rendu-Weber syndrome 448.0-(0)
 Osterreicher-Turner syndrome 756.88(2)
 Ostium primum 745.60(0)
 Ostium secundum 745.51(0)
 Ostrum-Furst syndrome 756.58(0)
 Otopalatodigital syndrome 759.84(5)

 Papillon-Leage and Psaume syndrome 759.89(3)
 Partial X deletion syndrome 758.61(1)
 Patau's syndrome 758.19(0)
 Pelizaeus-Merzbacher disease 330.0-(1)
 Pendred's syndrome 243.90(0)
 Penta X 758.88(1)

Peutz-Jeghers syndrome 759.60(0)
 Pierre Robin syndrome 756.03(0)
 Poland's syndrome 756.80(0)
 Polycystic disease (liver and kidney) 759.89(1)
 Polycystic renal disease or syndrome 753.13(0)
 Polycystic syndrome 759.89(1)
 Pompe's disease 271.01(0)
 Popliteal web syndrome 756.88(5)
 Potter's facies 754.01(0)
 Potter's syndrome 753.00(0)
 Pott's disease 015.0-(0)
 Prader-Willi syndrome 759.87(2)
 Pringle's disease or syndrome 759.5-(1)
 Progeria 259.80(0)
 Prune belly syndrome 756.72(0)
 Pseudo-Turner syndrome 758.69(2)

 Quinke's oedema 995.1-(0)

 Ramsey-Hunt syndrome 053.1-(0)
 Ranula 750.22(0)
 Rathke's pouch, cyst 253.80(0)
 Raynaud's syndrome 443.0-(0)
 Refsum's disease or syndrome 356.3-(0)
 Reiger's anomaly 743.48(0)
 Reiter's conjunctivitis 099.3-(1)
 Reiter's syndrome 099.3-(0)
 Rendu-Osler-Weber syndrome 448.0-(1)
 Reye's disease 331.8-(0)
 Riley-Day syndrome 742.81(0)

Ring chromosome 758.61(2)
 Ritter's disease 695.81(0)
 Robin's syndrome 756.03(0)
 Rocker bottom feet 755.61(0)
 Roger's disease 745.40(0)
 Rothmund's syndrome 757.30(3)
 Rothmund-Thomson disease or syndrome 757.30(3)
 Rotor's syndrome 277.43(0)
 Roussy-Levy syndrome 334.3-(1)
 Rubella syndrome 771.0-(0)
 Rubenstein-Taybi syndrome 759.84(1)
 Russell-Silver syndrome 759.82(3)

 Sanfilippo's syndrome 277.53(0)
 Scheie's syndrome 277.51(1)
 Scheuermann's disease 732.0-(0)
 Schilder's disease 341.1-(0)
 Schwartz's syndrome 756.88(0)
 Seckel's syndrome 759.82(7)
 Sex chromosome mosaic 758.88(0)
 Sezary's disease 202.2-(0)
 Short bowel syndrome 751.58(0)
 Sieman's syndrome 757.30(4)
 Silver's syndrome 759.82(3)
 Sirenomelia 759.84(4)
 Sjogren-Larsson syndrome 757.12(0)
 Sjogren's disease 710.2-(0)
 Smith-Lemli-Opitz syndrome 759.82(8)
 Soto's syndrome 756.88(3)
 Speilmeyer-Vogt disease 330.1-(2)

Sprengel's deformity 755.53(0)
 Stein-Leventhal syndrome 256.4-(0)
 Sternberg-McCune syndrome 756.51(1)
 Stevens-Johnson syndrome 695.12(0)
 Stilling's syndrome 378.7-(1)
 Still's disease 714.3-(0)
 Straight back syndrome 756.18(1)
 Stuart-Brass syndrome 453.0-(1)
 Sturge-Kalischer-Weber syndrome 759.61(2)
 Sturge-Weber(-Dimitri) syndrome 759.61(1)
 Sudden infant death syndrome 798.0-(0)
 Sydenham's chorea 392.9-(0)
 Sylvius' aqueduct anomaly 742.30(0)

 Taenzer's disease 757.43(0)
 Takayasu's arteritis, disease or syndrome 446.7-(0)
 Tangier disease 272.5-(0)
 Taussig-Bing complex 745.11(0)
 Tay-Sach's disease 330.1-(3)
 Telangiectatic pigmentation cataract 757.30(7)
 Thanatotropic dwarf 756.44(3)
 Thomson's disease or syndrome 757.30(2)
 Tietze's disease 733.6-(0)
 Todd's paralysis 344.80(0)
 Treacher-Collins syndrome 756.04(1)
 Triad disease or syndrome 759.34(1)
 Triple X female syndrome 758.85(0)
 Trisomy 13 syndrome 758.10(0)
 Trisomy 18 syndrome 758.20(0)
 Trisomy 21 syndrome 758.00(0)

Trisomy D NOS syndrome 758.11(0)
 Trisomy E NOS syndrome 758.21(0)
 Trisomy G syndrome 758.01(0)
 Trisomy syndrome NOS 758.52(0)
 Tuberculous Addison's disease 017.6-(0)
 Turck's syndrome 378.7-(2)
 Turner's karyotype 45 758.60(2)
 Turner's phenotype, isochromosome 758.61(3)
 Turner's phenotype, variant karyotype 758.61(4)
 Turner's phenotype, XO/XX mosaic 758.61(5)
 Turner's syndrome 758.69(1)
 Turner's syndrome, (XO) karyotype 758.60(1)

 Uhl's disease 746.86(0)
 Ullrich-Feichtiger syndrome 759.89(0)
 Ullrich(-Bonnevie)-Turner syndrome 758.69(0)
 Unverricht-Lundborg disease 333.22(0)
 Usher-Senear disease 694.4-(0)
 Usher's syndrome 759.89(4)

 Valsalva, sinus of, anomalies 747.24(0)
 Van der Hoeve's syndrome 756.50(4)
 V A T E R syndrome 759.89(5)
 Vincent's angina 101.--(0)
 Vincent's stomatitis 101.--(0)
 Vogt's disease 333.70(0)
 Von Gierke's disease 271.00(0)
 Von Hippel(-Lindau) syndrome 101.--(0)
 Von Recklinghausen-Applebaum disease 275.02(0)
 Von Recklinghausen's disease 237.7-(0)

Von Willebrand's disease 286.4-(0)

Vrolik's disease 756.50(5)

Vulcan ear(s) 744.23(0)

Waardenburg's syndrome 759.80(4)

Waterhouse-Friderichsen's syndrome 036.3-(0)

Wegener's granulomatosis 446.4-(0)

Weil's disease 100.0-(0)

Wenkebach's phenomenon 426.1-(0)

Werdnig-Hoffman disease 335.0-(0)

Werner's disease or syndrome 259.83(0)

Wharton's duct, anomaly or displacement of 750.23(0)

Whipple's disease 040.2-(0)

Whistling face syndrome 759.80(3)

Wildervanck's syndrome 756.11(1)

Wilm's tumour 189.0-(0)

Wilson-Mikity syndrome 770.70(0)

Wilson's disease 275.1-(0)

Wiskott-Aldrich disease or syndrome 279.11(0)

Wolff-Hirschorn syndrome 758.32(0)

Wolffian cyst 752.87(1)

Wolff-Parkinson-White syndrome 426.7-(0)

Wolman's disease 272.7-(4)

XO mosaic (Turner's phenotype) syndrome 758.61(0)

XXX female osteodystrophy 758.85(1)

XXY syndrome 758.70(0)

Zellweger's disease or syndrome 759.87(5)

Zollinger-Ellison syndrome 251.5-(0)