

**Table 1. List of conditions to be coded in the syndrome field**

	<b>ICD10-BPA</b>	<b>ICD9-BPA</b>	<b>OMIM</b>	<b>ORPHA</b>
<b>Syndromes – monogenic or unknown etiology</b>				
Aarskog syndrome	Q8710	75989	305400, 100050	915
Acrocephalopolysyndactyly (all types)	Q8700	755501	201000, 201020, 101120	65759, 65798,
Alagille syndrome	Q4471	75167	118450, 610205	52, 26100, 261619, 261629
Alport syndrome	Q8780	759870	104200, 203780, 301050	63, 88918, 88919, 88917
Angelman syndrome	Q8785	759899	105830	72
Aniridia-Wilms tumor syndrome WAGR	Q131	74342	194072, 612469	893
Apert syndrome (acrocephalosyndactyly)	Q8701	755500/02	101200	87
Bardet-Biedl syndrome	Q8781	759820	209900	110
Beckwith-Wiedemann syndrome (EMG syndrome)	Q8730	759874	130650	116
Blepharophimosis-ptosis syndrome	Q100	74360	110100	126
Branchiootorenal syndrome (BOR syndrome) CHARGE syndrome	Q178	75989	113650, 610896	107
Cleidocranial dysplasia (dysostosis)	Q300	75989	214800	138
Cockayne syndrome	Q7402	755551	119600, 216330	1452
	Q8711	759826	133540, 214150, 216400, 216411, 278780, 610756, 610758, 616570	191, 90321, 90322, 90324
Cornelia de Lange syndrome (de Lange syndrome)	Q8712	759821	122470, 300590, 300882, 610759, 614701	199
Crouzon syndrome	Q751	75601	123500	207
Dubowitz syndrome	Q8713	75989	223370	235
Ehlers-Danlos syndrome	Q796	75685	130000 + several	287 + several
Fragile X syndrome	Q992	75983	300624	908
Fraser syndrome (cryptophthalmos-syndactyly)	Q8702	759892	219000	2052
Frontonasal dysplasia	Q7581	75400	136760	391474
Gardner syndrome	Q8583	759630	175100	79665
Gorlin-Chaudhry-Moss syndrome	Q878	759898	233500	2095
Hallermann-Streiff syndrome	Q8705	756050	234100	2108
Holt-Oram syndrome (heart-hand syndrome)	Q8720	759842	142900	392
Hypoglossia-Hypodactylia syndrome	Q878	759846	103300	989
Incontinentia pigmenti	Q823	75735	308300	464
Ivemark syndrome	Q206	75989	208530	97548
Kartagener syndrome	Q8934	759340	244400	98861
Kaufman-McKusick syndrome	Q518	75248	236700	2473
Klippel-Feil syndrome	Q761	756110	118100, 214300, 613702	2345
Klippel-Trenaunay (-Weber) syndrome (angioosteohypertrophy) Larsen syndrome	Q8721	759840	149000, 608355	2346
Laurence-Moon syndrome	Q7484	75581	150250, 245600	503, 284139
Lenz microphthalmos syndrome	Q8781	759822	245800	2377
Marfan syndrome	Q112	74310	309800	568
McCune-Albright syndrome	Q874	759860	154700, 610168	558
	Q781	756512	174800	562

Meckel Gruber syndrome	Q6190	75989	249000 + several	564
Melnick-Needles syndrome	Q778	759845	309350	2484
Miller-Dieker syndrome	Q043	74224	247200	531
Noonan syndrome	Q8714	759896	163950 + several	648
Orofacialdigital syndrome type 1	Q8707	759802	311200	2750
Orofacialdigital syndrome type 2	Q8707	759802	252100	2751
Otopalatodigital syndrome type 1	Q870F	759845	311300	669, 90650
Otopalatodigital syndrome type 2	Q870F	759845	304120	669, 90652
Pena-Shokeir syndrome	Q870E	75989	208150, 300073	994
Peutz-Jeghers syndrome	Q8580	759600	175200	2869
Pfeiffer syndrome	Q750	755501	101600	710
Poland syndrome	Q7982	75680	173800	2911
Popliteal pterygium syndrome	Q798	756885	119500, 263650	1300, 1234
Prader-Willi syndrome	Q8715	759872	176270, 615547	739
Robinow-Silverman-Smith syndrome	Q8716	75989	180700, 268310	97360, 3107, 1507
Rothmund-Thomson syndrome	Q828	757303	268400	2909
Rubinstein-Taybi syndrome	Q8723	759841	180849, 610543, 613684	783, 353281, 353284
Seckel syndrome	Q8718	759827	210600 + several	808
Silver-Russel syndrome	Q8717	759823	180860, 312780	813
Sjögren-Larsson syndrome	Q871A	75712	270200	816
Smith-Lemli-Opitz syndrome	Q87.19	759828	270400	818
Smith-Magenis syndrome	Q878	75989	182290	819
Sotos syndrome	Q8731	756883	117550, 617169	821
Stickler syndrome	Q8709	75989	108300, 604841, 184840	828, 90653, 90654, 166100
Sturge-Weber syndrome	Q8581	759611	185300	3205
TAR syndrome	Q8725	75526	274000	3320
Treacher-Collins syndrome (mandibulofacial dysostosis)	Q870A	756041	154500, 248390, 613717	861
Trichorhinophalangeal syndrome	Q870B	75989	190350, 190351	77258
Van der Woude syndrome	Q380	75989	119300, 604547, 606713	888
Velocardiofacial syndrome/DiGeorge syndrome	D82.1	27910	188400, 192430	567
Von Hippel Lindau syndrome	Q8582	75962	193300	892
Waardenburg syndrome	E7030	759804	193500 + several	3440
Weaver syndrome	Q8732	75989	277590	3447
Weill Marchesani syndrome	Q875	75989	277600, 608328, 614819	3449
Whistling face syndrome (Freeman-Sheldon syndrome)	Q870C	759807	193700, 277720, 616266	2053
Williams syndrome	Q8784	75989	194050	904
Zellweger syndrome	Q8783	759875	214100 + several	912
<b>Teratogenic syndromes</b>				
Fetal alcohol syndrome	Q860	76076	-	1915
Fetal cytomegalovirus (CMV) syndrome	P351	77110	-	294
Fetal hydantoin syndrome	Q861	76070	-	1912
Fetal rubella syndrome	P350	77100	-	290
Fetal thalidomide syndrome	Q8682	7607	-	3312
Fetal toxoplasmosis	P371	77121	-	858
Fetal valproate syndrome	Q868	7607	-	1906
Fetal warfarin syndrome	Q862	7607	-	1914
Fetal zika syndrome	P358	-	-	448237

**Skeletal dysplasias**

Achondrogenesis type 1A and type 1B	Q7700	75644	200600, 600972	932, 93299, 93298
Achondrogenesis type 2 (incl. hypochondrogenesis)	Q7701/ 02	75644	200610	93296
Achondroplasia/hypochondroplasia	Q774	75643	100800	15
Acrodysostosis	Q778	75658	101800	950
Albers-Schönberg syndrome (osteopetrosis)	Q782	756540	166600	53
Camurati-Engelmann disease (diaphyseal dysplasia)	Q783	756551	131300	1328
Chondrodysplasia punctata, rhizomelic, type 1	Q773	75644	215100	177
Chondrodysplasia punctata 2, X-linked	Q773	75644	302960	35173
Diastrophic dysplasia	Q775	756441	222600	628
Ellis van Creveld syndrome	Q776	75652	225500, 617088	289
Enchondromatosis (including Maffucci)	Q784/ Q7840	75641/75642	166000, 614569	296, 163634
Jeune asphyxiating thoracic dystrophy	Q772	75640	208500 + several	474
Kniest dysplasia	Q778	75658	156550	485
Metaphyseal chondrodysplasia	Q7781	75645	156400	33067
Metaphyseal dysplasia, Pyle syndrome	Q785	75655	265900	3005
Metatropic dysplasia	Q7780	756442	156530	2635
Multiple congenital exostosis, diaphyseal aclasia	Q786	75657	133700, 133701, 600209	321,
Nail patella syndrome (onycho-osteodysplasia)	Q8722	75683	161200	2614
Osteogenesis imperfecta	Q7800	75650	166200, 166210, 166220, 166230, 259420, 259440, 610682, 610915, 610967, 610968, 613848, 613849, 613982, 614856, 615066, 615220, 616229, 616507	666, 216796, 216804, 216812, 216820, 216828
Osteopoikilosis	Q7880	75656	166700	1306
Spondyloepiphyseal dysplasia congenita	Q777	75646	183900	94068
Spondyloepiphyseal dysplasia tarda	Q777	75646	313400	93284
Thanatophoric dysplasia	Q771	756443	187600, 187601	2655, 1860, 93274

**Associations**

MURCS syndrome	Q518	75249	601076	2578
Oculoauriculovertebral spectrum (Goldenhar syn)	Q8704	75606	164210	374
VATER association	Q8726	759895	192350	887

**Table 2. List of conditions which should not be coded as syndromes (code as malformation)**

	<b>ICD10- BPA</b>	<b>ICD9- BPA</b>	<b>OMIM</b>	<b>ORPHA</b>
<b>Sequences</b>				
Amniotic band sequence	Q7980	75688	217100	1034
Caudal regression sequence	Q8980	75989	600145	3027
Möebius sequence	Q8706	3568	157900	570
Pierre-Robin sequence	Q8708	75603	261800	718
Potter sequence	Q606	753000	191830	1848
Prune-belly sequence	Q794	75672	100100	2970
Sirenomelia	Q8724	759844	600145	3169
<b>Malformations with syndrome names but not a syndrome</b>				
Apple peel syndrome	Q411	75111	-	-
Arnold-Chiari syndrome	Q070	74288	118420, 207950	268882, 1136
Arthrogryposis multiplex congenita	Q743	75688	208100	1037
Congenital blind loop syndrome	Q438	75119	-	-
Conjoined twins	Q894	75949	-	-
Constriction ring syndrome of upper limb NOS	Q719	75558	217100	295000
Constriction ring syndrome of lower limb NOS	Q729	75568	217100	295000
Cryptophthalmos syndrome (isolated cryptophthalmos)	Q112	74380	123570	91396
Cyclops syndrome	Q8703	75989	-	-
Dandy-Walker syndrome/malformation	Q031	742.31	220200	217
Eisenmenger syndrome	Q2181	745.41	-	97214
Herlitz syndrome (epidermolysis bullosa)	Q811	757.33	226700	79404
Hirschsprung syndrome/disease	Q431	751.30	142623	388
Hypoplastic left heart syndrome	Q234	746.70	241550, 614435	2248
Hypoplastic right heart syndrome	Q226	74688	-	98723
Lutembacher syndrome (ASD+mitral stenosis)	Q2114	74552	-	-
Marcus Gunn syndrome/phenomenon	Q0780	74280	154600	91412
Megacystis-megaureter syndrome/sequence	Q6476	75322	-	238637
Pharyngeal pouch syndrome	D821	27910	188400	567
Scimitar syndrome	Q268	74743	608281	185
Siemen syndrome	Q828	75735	305100	181
Small left colon syndrome	Q432	75152	-	-
Taussig-Bing syndrome	Q201	74511	-	99045