

ESPE Code	Diagnosis of syndromes with endocrine features according to the European society of Pediatric Endocrinology (ESPE)	OMIM	ICD-10
14A	CHROMOSOMAL ABNORMALITIES (deletions or duplications of complete or half chromosomes)		
14A.1	Chromosome 18q deletion syndrome <i>If associated with short stature: also classify as 1A.1a</i>	#601808	Q93.5
14A.2	Down syndrome, trisomy 21 <i>If associated with any other disorder, also classify there For example: If associated with short stature: also classify as 1A.1a If associated with obesity: also classify as 5B.3a If associated with autoimmune hypothyroidism: also classify as 7A.3b If associated with diabetes mellitus: also classify as 11A.3h.1</i>	#190685	Q90
14A.3	Klinefelter syndrome (47,XXY) <i>If associated with tall stature: also classify as 2A.1a If associated with obesity: also classify as 5B.3a If associated with hypogonadism: also classify as 9A.0 If associated with diabetes mellitus: also classify as 11A.3h.2</i>		Q98.0
14A.4	Mixed gonadal dysgenesis (45,X/46,XY) <i>If associated with disorder of sex development, primarily classify as 4A.1 If associated with short stature: also classify as 1A.1a</i>		Q99.9
14A.5	Turner syndrome (Ulrich-Turner syndrome) (45,X or variants) <i>If associated with short stature: also classify as 1A.1a If associated with obesity: also classify as 5B.3a If associated with gonadal dysgenesis: also classify as 10A.1a If associated with diabetes mellitus: also classify as 11A.3h.3</i>		Q96.9
14A.6	YYY syndrome <i>If associated with tall stature: also classify as 2A.1a</i>		Q98.5
14B	CONGENITAL DYSMORPHIC SYNDROMES		
14B.1	Aarskog-Scott syndrome <i>If associated with short stature: also classify as 1A.1a If associated with shawl scrotum: also classify as 9F.2</i>	100050	Q87.1
14B.2	Albright's hereditary osteodystrophy, Pseudohypoparathyroidism <i>If associated with hypocalcaemia: also classify as 12B.5a If associated with short stature: also classify as 1A.3y If associated with obesity: also classify as 5B.2a</i>	#103580	E20.1
14B.3	Alström syndrome <i>If associated with obesity: also classify as 5A.2a If associated with diabetes mellitus: also classify as 11A.3h.11</i>	#203800	Q87.8
14B.4	Bannayan-Riley-Ruvalcaba syndrome <i>If associated with tall stature: also classify as 2A.3a</i>	#153480	Q87.3
14B.5	Beckwith-Wiedemann syndrome <i>If associated with tall length/stature: also classify as 2A.4a If associated with hypoglycaemia: also classify as 11B.1a.5</i>	#130650	Q87.3
14B.6	Bloom syndrome <i>If associated with short stature: also classify as 1A.1a</i>	#210900	Q82.8
14B.7	Carpenter syndrome <i>If associated with obesity: also classify as 5B.2a</i>	201000%	Q87.0
14B.8	Cohen syndrome <i>If associated with obesity: also classify as 5B.2a</i>	#216550	Q87.8
14B.9	Cornelia de Lange syndrome <i>If associated with short stature: also classify as 1A.1a</i>	#122470 #300590 #610759	Q87.1

14B.10	DiGeorge/velocardiofacial syndrome syndrome (part of CATCH 22 complex) <i>If associated with short stature: also classify as 1A.1a If associated with hypocalcaemia: also classify as 12B.2a</i>	#188400	Q87.1
14B.11	Ehlers-Danlos syndrome <i>If associated with osteoporosis: also classify as 12D.1a</i>	#130000 (to 80) #225400 %229200 %305200	Q79.6
14B.12	Elejalde syndrome <i>If associated with tall stature: also classify as 2A.3a</i>	200995	Q87.3
14B.13	Fanconi renotubular syndrome <i>If associated with short stature: also classify as 1B.3a.0 If associated with rickets: also classify as 12C.2a</i>	134600%	E72.0
14B.14	Fragile X syndrome <i>If associated with tall stature: also classify as 2A.3a</i>	#300624	Q99.2
14B.15	Gitelman syndrome <i>If associated with hypocalcaemia: classify as 12B.6e</i>	#263800	
14B.16	Kallmann syndrome <i>Primarily classified as 6A.3a.1 If associated with tall stature: also classify as 2B.6a</i>	+308700 #147950 #244200	E23.0
14B.17	Klippel-Trenaunay-Weber syndrome <i>If associated with tall stature: also classify as 2A.4a</i>	149000%	Q87.2
14B.18	Laurence-Moon-Bardet-Biedl syndrome <i>If associated with obesity: also classify as 5B.2a If associated with hypogonadism: also classify as 9A.0 If associated with diabetes mellitus: also classify as 11A.3h.7</i>	245800 #209900	Q87.8
14B.19	Leri-Weill dyschondrosteosis <i>If associated with short stature: also classify as 1A.1a</i>	#127300	Q77.8
14B.20	Marfan syndrome <i>If associated with tall stature: also classify as 2A.2a If associated with osteoporosis: also classify as 12D.1a</i>	#154700	Q87.4
14B.21	Marshall-Smith syndrome <i>If associated with tall stature: also classify as 2A.3a</i>	602535	
14B.22	McCune-Albright syndrome <i>If associated with tall stature: also classify as 2B.1b If associated with pseudoprecocious puberty: also classify as 3A.2c.0 If associated with hyperthyroidism: also classify as 7B.1d.1 If associated with phosphogenic rickets: also classify as 12C.2a</i>	#174800	Q87.1
14B.23	Nevo syndrome <i>If associated with tall stature: also classify as 2A.3a</i>	#601451	E87.3
14B.24	Noonan syndrome <i>If associated with short stature: also classify as 1A.1a If associated with hypogonadism: also classify as 9A.0</i>	#163950	Q87.1
14B.25	Prader-Willi(-Labhart) syndrome <i>If associated with short stature: also classify as 1A.1a If associated with obesity: also classify as 5B.2a If associated with hypogonadotrophic hypogonadism: also classify as 6A.3d If associated with hypothalamic-pituitary disorders: also classify as 6E.2a If associated with diabetes mellitus: also classify as 11A.3h.10</i>	#176270	Q87.1
14B.26	Proteus syndrome <i>If associated with tall stature: also classify as 2A.4a</i>	#176920	Q87.3
14B.27	Von Recklinghausen's disease (neurofibromatosis type 1) <i>If associated with short stature: also classify as 1A.1a If associated with precocious puberty: also classify as 3A.1a and 6F.1a.5</i>	162200	Q85.0
14B.28	Rieger syndrome <i>If associated with growth hormone deficiency: also classify as 1B.3a.0 If associated with ACTH deficiency: also classify as 6A.1 If associated with TSH deficiency: also classify as 6A.2 If associated with gonadotrophin deficiency: also classify as 6A.3d</i>	#180500	Q87.1
14B.29	Rubinstein-Taybi syndrome <i>If associated with short stature: also classify as 1A.1a</i>	#180849	Q87.2

14B.30	Septo-optic dysplasia <i>Primarily classified as 6E.1a If associated with short stature: also classify as 1B.3a.3 If associated with diabetes insipidus: also classify as 13A.1b.1</i>	#182230	Q04.4
14B.31	Silver-Russell syndrome <i>If associated with short stature: also classify as 1A.1a If associated with precocious or delayed puberty: also classify according to the codes in chapter 3</i>	180860%	Q87.1
14B.32	Simpson-Golabi-Behmel syndrome (mutations in GPC3) <i>If associated with tall stature: also classify as 2A.3a</i>	#312870	Q87.3
14B.33	Smith-Lemli-Opitz syndrome <i>If associated with a disorder of sex development: also classify as 4B.2a If associated with low cholesterol level: also classify as 11C.4a</i>	#270400	Q87.1
14B.34	Sotos syndrome <i>If associated with tall stature: also classify as 2A.3a</i>	#117550	Q87.3
14B.35	Steinert's disease, myotonic dystrophy 1 <i>If associated with precocious puberty: also classify as 3A.1y If associated with delayed puberty: also classify as 3D.0 If associated with hypothyroidism: also classify as 7A.3y If associated with hyperthyroidism: also classify as 7B.2a If associated with adrenal insufficiency: also classify as 8A.3a If associated with hypogonadism: also classify as 9A.0 If associated with diabetes mellitus: also classify as 11A.3h.8</i>	#160900	G71.1
14B.36	Weaver syndrome <i>If associated with tall stature: also classify as 2A.3a</i>	#277590	Q87.3
14B.37	Williams-Beuren syndrome <i>If associated with short stature: also classify as 1A.1a If associated with hypercalcaemia: also classify as 12E.4a</i>	#194050	Q87.1
14B.38	Wolfram syndrome (DIDMOAD) <i>If associated with diabetes mellitus: also classify as 11A.3h.4 If associated with diabetes insipidus: also classify as 13A.1a.2</i>	#222300	E10 E23.2