

Klinik für Kinder- und Jugendmedizin, Sektion Pädiatrische Endokrinologie und Diabetologie
Sektionsleiter Prof. Dr. M. Wabitsch

ESPE Code	<i>Diagnosis of disorders of bone metabolism including calcium and phosphate metabolism according to the European society of Pediatric Endocrinology (ESPE)</i>	OMIM	ICD-10
12A	TRANSIENT HYPOCALCAEMI A (NEONATAL)		E83.5
12A.1	Early neonatal		P71.1
12A.1a	Prematurity		
12A.1b	Asphyxia		
12A.1c	Infant of diabetic mother		
12A.1d	Perinatal stress or trauma		
12A.2	Late neonatal		P71.1
12A.2a	Disorders classified elsewhere: <i>Conditions classified under permanent hypocalcaemia, hypomagnesaemia</i>		
12A.2b	High milk phosphate load		P71.0, P71.1
12A.2c	Parenteral nutrition		
12A.2d	Exchange transfusions		
12A.2e	Chronic alkalosis or bicarbonate treatment		
12A.2f	Maternal hypercalcaemia		
12A.2g	Maternal vitamin D deficiency		
12A.2h	Transient hypoparathyroidism		P71.4

12B	PERMANENT HYPOCALCAEMIA		
12B.1	Genetic disorders of the calcium-sensing receptor		
12B.1a	Autosomal-dominant hypocalcaemia	#146200	
12B.1b	Autosomal-dominant hypocalcaemia with Bartter-like features	601199	
12B.2	Hypoparathyroidism		E20.9
12B.2a	Disorders classified elsewhere: <i>DiGeorge syndrome types 1 and 2</i> (14B.10) <i>Autoimmune polyglandular syndrome type 1 (APECED syndrome)</i> (14C.4a)		E20.8
12B.2b	X-linked hypoparathyroidism	307700%	E20.8
12B.2c	Mitochondrial disorders		
12B.2c.1	Kearns-Sayre	#530000	
12B.2c.2	MELAS	#540000	
12B.2c.3	Pearson marrow-pancreas syndrome	#557000	
12B.2c.4	tRNA-Leu mutation	*590050	
12B.2c.9	Other		
12B.2d	Hypoparathyroidism, deafness and renal anomalies (Barakat syndrome)	#146255	
12B.2e	Long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD deficiency) [primary 11B.5d]	*600890	
12B.2f	Other familial syndromes		E20.8
12B.2f.1	Kenny-Caffey syndrome type 1	#244460	
12B.2f.2	Sanjad-Sakati	#241410	
12B.2f.9	Other specified syndromes		
12B.2g	Acquired hypoparathyroidism		
12B.2g.1	Parathyroid surgery		
12B.2g.2	Isolated autoimmune hypoparathyroidism		
12B.2g.3	Iron overload		
12B.2g.4	Other specified acquired forms		
12B.2z	Idiopathic		E20.0
12B.3	PTH defects (autosomal-dominant or autosomal-recessive familial isolated hypoparathyroidism)	#146200	E20.8
12B.4	PTH/PTHrP receptor defects		
12B.4a	Pseudohypoparathyroidism type 1b	#603233	
12B.4z	Other PTH/PTHrP receptor defects, unspecified		
12B.5	Post-receptor defects		
12B.5a	Classified elsewhere: <i>Pseudohypoparathyroidism type Ia (Albright's hereditary osteodystrophy, 14B.2)</i>		E20.1
12B.5b	Pseudohypoparathyroidism type Ic	#103580	E20.1
12B.5c	Pseudohypoparathyroidism type II	203330%	E20.1
12B.5d	Pseudohypoparathyroidism with testotoxicosis	#176410	E20.1
12B.6	Magnesium deficiency		E83.4
12B.6a	Familial primary hypomagnesaemia	#248250	
12B.6b	Familial hypomagnesaemia with hypercalciuria, nephrocalcinosis, and severe ocular involvement	#248190	
12B.6c	Isolated renal magnesium wasting	#154020	
12B.6d	Hypomagnesaemia with secondary hypocalcemia	#602014	
12B.6e	Gitelman syndrome [primary 14B.15]	#263800	
12B.7	Calciopenic rickets (see 12C.1)		

12B.8	Systemic conditions associated with hypocalcaemia		
12B.8a	Tumour lysis syndrome		
12B.8b	Renal osteodystrophy		N25.0
12B.8c	AIDS		
12B.8y	Other specified conditions		
12B.8z	Other conditions, unspecified		
12C	RICKETS		
12C.1	Calciopenic rickets		E55.0
12C.1a	Nutritional (vitamin D deficiency)		
12C.1a.1	Malabsorption		
12C.1a.2	Liver disease		
12C.1a.3	Anticonvulsant treatment (phenobarbital, phenytoin)		
12C.1a.4	Renal osteodystrophy		N25.0
12C.1a.5	Calcium deficiency rickets		
12C.1b	Genetic		
12C.1b.1	Vitamin D 1 _α -hydroxylase deficiency (formerly known as pseudo-vitamin D-deficiency rickets, vitamin D dependent rickets type I)	#264700	E83.3
12C.1b.2	Hereditary 1,25(OH) ₂ D-resistant rickets (formerly known as pseudo-vitamin D-deficiency rickets type II, vitamin D-dependent rickets type II, calcitriol-resistant rickets)	#277400	E83.3
12C.2	Phosphopenic rickets		E83.3
12C.2a	Classified elsewhere: <i>McCune-Albright syndrome</i> (14B.22) <i>Renal tubular disorders – Fanconi renal tubular syndrome</i> (14B.13)		
12C.2b	Familial hypophosphataemic rickets		
12C.2b.1	X-linked hypophosphataemic rickets	#307800	
12C.2b.2	Autosomal-dominant hypophosphataemic rickets	#193100	
12C.2b.3	Hereditary hypophosphataemic rickets with hypercalciuria	#241530	
12C.2b.4	Hypophosphataemic nonrachitic bone disease	146350%	
12C.2c	Tumour-induced osteomalacia		
12C.2d	Decreased phosphate intake		
12C.3	Hypophosphatasia (if not associated with rickets, classify under 12E.5l)	#146300 #241500	E83.3
12D	OSTEOPOROSIS		M81.9
12D.1	Genetic defects		
12D.1a	Classified elsewhere: <i>Ehlers-Danlos syndrome</i> (14B.11) <i>Marfan syndrome</i> [primary 14B.20, other secondary 2A.2a]		
12D.1b	Osteogenesis imperfecta (types I–VII)	#166200 #166210 #166220 #166240 #259420	
12D.1c	Homocystinuria [if tall, classify also as 2A.2c]	236200	
12D.1d	Other genetic defects		
12D.2	Chromosomal defects		M82.8
12D.3	Endocrine disorders		M82.0
12D.4	Iatrogenic causes		M81.4
12D.5	Nutritional disorders		M81.3
12D.6	Chronic diseases		M82.8
12D.7	Malignancies		M82.8

12D.8	Disuse		M81.2
12D.9	Other specified disorders, e.g. muscle and neuromuscular disorders	M81.8	
12D.10	Idiopathic juvenile osteoporosis		M81.5
12E	HYPERCALCAEMI A		E83.5
12E.1	Disorders of the calcium-sensing receptor		
12E.1a	Familial benign hypercalcaemia (familial hypocalciuric hypercalcaemia)	#145980	
12E.1b	Neonatal severe primary hyperparathyroidism	#239200	
12E.1c	Calcium-sensing receptor blocking antibodies		
12E.2	Disorders of the parathyroid glands/PTH oversecretion		E21
12E.2a	Disorders classified elsewhere: <i>Multiple endocrine neoplasia type 1</i> (14C.5a) <i>Multiple endocrine neoplasia type 2a</i> (14C.5b) <i>Multiple endocrine neoplasia type 2b</i> (14C.5c)		
12E.2b	Familial isolated hyperparathyroidism, type 1	#145000	E21.0
12E.2c	Familial isolated hyperparathyroidism, type 2 (jaw tumour)	#145001	E21.0
12E.2d	Sporadic parathyroid adenomas		E21.4
12E.2e	Parathyroid carcinomas		E21.4
12E.2f	Tertiary hyperparathyroidism		E21.4
12E.2y	Other PTH abnormalities (specified)		E21.4
12E.3	PTH/PTHrP Receptor abnormalities		
12E.3a	Metaphyseal chondrodysplasia, Jansen type	#156400	Q78.9
12E.4y	Other PTH/PTHrP receptor abnormalities (specified)		
12E.4	Abnormal vitamin D metabolism		
12E.4a	Disorders classified elsewhere: <i>Williams-Beuren syndrome</i> (14B.37)		
12E.4b	Idiopathic infantile hypercalcaemia	#19405	
12E.5	Miscellaneous causes of hypercalcaemia in childhood		E83.5
12E.5a	Vitamin D (or vitamin D metabolite) intoxication		E67.3
12E.5b	Sarcoidosis, other granulomatous diseases		D86.9
12E.5c	Immobilisation		E83.5
12E.5d	Hypercalcaemia of malignancy		E83.5
12E.5e	Subcutaneous fat necrosis		
12E.5f	Excessive calcium supplementation		
12E.5g	Congenital lactase deficiency	#223000	
12E.5h	Disaccharidase deficiency	#222900	
12E.5i	Endocrine forms (adrenal insufficiency, severe congenital hypothyroidism, thyrotoxicosis) (<i>primarily classified elsewhere</i>)		
12E.5j	Down syndrome	#190685	Q90
12E.5k	AIDS		B24
12E.5l	IMAGe syndrome	300290	
12E.5m	Hypophosphatasia	#241500	E83.3
12E.5y	Other specified disorders		
12E.5z	Other disorders (unspecified)		