ESPE Code	***Diagnosis of growth disorders according to the European Society of Pediatric Endocrinology (ESPE)***	**OMIM**	**ICD-10**
1	**SHORT STATURE**		
1A	**PRIMARY GROWTH FAI LURE**		
1A.1	**Clinically defined syndromes**		
1A.1a	Syndromes classified elsewhere:		
	45,X/46,XY disorder of sex development (4A.1)		
	45,X and variants with female phenotype ((Ulrich)-Turner		
	syndrome) (14A.5)		
	Phenotypic male with X/XY mosaicism (14A.4)		
	18q deletion syndrome (14A.1)		
	Aarskog-Scott syndrome (14B.1)		
	Bloom syndrome (14B.6)		
	Cornelia de Lange syndrome (14B.9)		
	DiGeorge syndrome (velocardiofacial syndrome) (14B.10)		
	Down syndrome (14A.2)		
	Kabuki makeup syndrome (14C.4f)		
	Noonan syndrome (14B.24)		
	Prader-Willi-Labhart syndrome (14B.25)		
	Von Recklinghausen's disease (neurofibromatosis type 1) (14B.27)		
	Rubinstein-Taybi syndrome (14B.29)		
	Silver-Russell syndrome (14B.31)		
	Williams-Beuren syndrome (14B.37)		
1A.1y	Other specified syndromes, e.g. Three M slender-boned		
	dwarfism, Seckel syndrome, Mulibrey nanism		
1A.1z	Other syndromes associated with short stature, unspecified		
1A.2	**Small for gestational age (SGA) with failure of catch-up growth**	**P05.1**	
1A.2a	Disorders classified elsewhere:		
	IGF-I deficiency (1B.4e)		
	IGF resistance (1B.4f)		
	1A.2y Due to known cause (specify), e.g. prenatal infection, drugs,		P00.2–4
	smoking, alcohol		
1A.2z	Idiopathic		

1A.3	**Skeletal dysplasias (constitutional disorders of bone)**		**Q77, Q78**
1A.3a	Achondroplasia group (group 1)		
1A.3a.1	Achondroplasia	#100800	
1A.3a.2	Hypochondroplasia	#146000	
1A.3a.8	Other specified disorders included in this group (thanatophoric dysplasia, SADDAN)		
1A.3b	Type II collagenopathies (COL2A1 defects) (group 8)		
1A.3b.1	Spondyloepiphyseal dysplasia congenita	#183900	
1A.3b.8	Other specified disorders in this group		
1A.3c	Mesomelic dysplasias (group 16)		
1A.3c.1	Dyschondrosteosis (Leri-Weill and other defects in the SHOX gene, e.g. in children without dysmorphic features) [primary: 14B.19]	#127300	
1A.3c.2	Langer type (homozygous dyschondrosteosis)	#249700	
1A.3c.8	Other specified disorders included in this group		
1A.3d	Dysostosis multiplex group (group 22)		
1A.3d.1	Mucopolysaccharidosis (type IH, IS, II–VII)		E76
1A.3d.2	Mucolipidosis (type II and III)	#252500	E77.0
		#252600	
1A.3d.8	Other specified disorders included in this group		
1A.3e	Dysplasias with decreased bone density (group 24)		
1A.3e.1	Osteogenesis imperfecta I–VI [primary: 12D.1b]		
1A.3e.8	Other specified disorders included in this group		
1A.3f	Dysplasias with defective mineralisation (group 25)		
1A.3f.1	Hypophosphatasia	#241500	
1A.3f.2	Hypophosphataemic rickets [primary 12C.2]	#307800	
1A.3f.8	Other specified disorders included in this group		
1A.3y	Disorders included in other groups (2–7, 9–15, 17–21, 23, 26–33)		
1A.3z	Other skeletal dysplasia, unspecified		
1B	**SECONDARY GROWTH FAI LURE**		
1B.1	**Insufficient nutrient intake (malnutrition)**		**E40–46**
1B.2	**Disorders in organ systems**		
1B.2a	Cardiac disorders		Q20–28
1B.2b	Pulmonary disorders, e.g. cystic fibrosis		J40–99
			(E84)
1B.2c	Liver disorders		K70–77
1B.2d	Intestinal disorders, e.g. Crohn´s disease, malabsorption syndromes, short bowel syndrome		K50–52 K90–93
1B.2e	Renal disorders, e.g. Fanconi syndrome, renal acidosis		N10–19
			N25–29
1B.2f	Chronic anaemia		D50–64
1B.2g	Multiorgan disorders		
1B.2h	Muscular and neurological disorders, e.g. Duchenne muscular dystrophy, congenital myotonia		G71–73
1B.2i	Connective tissue diseases, e.g. juvenile arthritis		M08
1B.2y	Other specified organ or systemic disorders		

1B.3	**Growth hormone deficiency (secondary IGF deficiency)**		**E23.0**
	In case of multiple pituitary deficiencies, classify the various deficiencies separately:		
	ACTH deficiency (6A.1)		
	TSH deficiency (6A.2)		
	Gonadotropin deficiency (6A.3)		
	Prolactin deficiency (6A.4)		
	Vasopressin deficiency (diabetes insipidus) (13A.1)		
1B.3a	Congenital growth hormone deficiency		E23.0
1B.3a.0	Disorders classified elsewhere:		
	Associated with complex syndromes: Fanconi renotubular syndrome (14B.13)		
	Rieger syndrome (14B.28)		
	Kabuki make-up syndrome (14C.4f)		
1B.3a.1	Associated with other complex syndromes: ectodactyly-ectodermal dysplasia clefting syndrome		E23.0
1B.3a.2	Known genetic defects		E23.0
1B.3a.2a	– HESX1	*601802	
1B.3a.2b	– LHX3	*600577	
1B.3a.2c	– LHX4	*602146	
1B.3a.2d	– PROP1	*601538	
1B.3a.2e	– POU1F1	*173110	
1B.3a.2f	– GHRHR	*139191	
1B.3a.2g	– GH	*139250	
1B.3a.2y	– Other specified genetic defects		
1B.3a.3	– Associated with cerebral or facial malformations, e.g. septooptic dysplasia [primary 6E.1a], empty sella syndrome, solitary central maxillary incisor syndrome, mid-line palateal cleft, arachnoid cyst, congenital hydrocephalus, hypoplastic anterior pituitary + missing stalk + ectopic posterior pituitary (HME)		Q04.4 Q37.9 O35.0
	Excluded: Known genetic defects (1B.3a.2)		
1B.3a.4	– Associated with prenatal infection, e.g. rubella		P35.0
1B.3a.8	– Associated with other specified disorders		E23.0
1B.3a.9	– Idiopathic		E23.0
1B.3a.9a	– 'Classical' idiopathic growth hormone deficiency		
1B.3a.9b	– Neurosecretory dysfunction7		
1B.3b	Acquired growth hormone deficiency		E23.0
1B.3b.1	– Craniopharyngioma		D44.4
1B.3b.2	– Other pituitary tumours, e.g. germinoma, hamartoma		M9064/3
1B.3b.3	– Cranial tumours distant from the pituitary/hypothalamic area, e.g. astrocytoma, ependymoma, glioma, medulloblastoma, nasopharyngeal tumour		C71 M9400/3 M9391/3 M9380/3 M9470/3
IB.3b.4	– Tumours outside the cranium, e.g. leukaemia, lymphoma		C91–96
IB.3b.5	– Head trauma		S06
IB.3b.6	– Central nervous system infection		G01–08
IB.3b.7	– Granulomatous diseases, e.g. histiocytosis		D76
IB.3b.8	– Vascular anomaly		Q28
IB.3b.9	– Other causes, unspecified		E23.0

1B.4	**Other disorders of the growth hormone-IGF axis (primary IGF-I deficiency and resistance)**		
1B.4a	Bio-inactive growth hormone (Kowarski syndrome)	#262650	E34.3
1B.4b	Abnormalities of the growth hormone receptor (growth hormone insensitivity syndrome, Laron syndrome)	#262500	E34.3
1B.4c	Abnormalities of GH signal transduction, e.g. STAT5B defect	#245590	E34.3
1B.4d	ALS (acid-labile subunit) deficiency	#601489	E34.3
1B.4e	IGF-I deficiency	#608747	E34.3
1B.4f	IGF resistance (IGF1R defects, postreceptor defects)	#270450	E34.3
1B.4z	Other disorders, unspecified		
1B.5	**Other endocrine disorders**		
1B.5a	Disorders classified elsewhere:		
	Cushing syndrome (8C.1)		
	Hypothyroidism (7A)		
	Leprechaunism (11A.3b.2)		
	Poorly controlled diabetes mellitus, Mauriac syndrome (14C.2)		
1B.5b	Short adult stature caused by accelerated bone maturation, e.g. precocious puberty (3A), hyperthyroidism (7B), congenital adrenal hyperplasia (8A.1), exogenous estrogens or androgens (3A.2d)		
1B.5y	Other specified disorders		E34.3
1B.6	**Metabolic disorders**		
1B.6a	Disorders classified elsewhere: Disorders of calcium and phosphorus metabolism (1A.3 and 12)		
1B.6b	Disorders of carbohydrate metabolism		E74
1B.6c	Disorders of lipid metabolism		E75
1B.6d	Disorders of protein metabolism		E70–72
1B.6y	Other specified metabolic disorders		E76–83
1B.6z	Other metabolic disorders, unspecified		E88
1B.7	**Psychosocial**		**E34.3**
1B.7a	Psychosocial (emotional) deprivation		T74
1B.7b	Anorexia nervosa		F50
1B.7c	*Depression*		F32.9
1B.7y	Other specified psychosocial causes		E34.3
1B.8	**Iatrogenic**		
1B.8a	Systemic glucocorticoid therapy (primary 8C.1c		T38.0
1B.8b	Local glucocorticoid therapy (inhalation, intestinal, other)		T49.0
1B.8c	Other medication		T36–50
1B.8d	Treatment of childhood malignancy		T66
1B.8d.1	Total body irradiation		T45.1
1B.8d.2	Chemotherapy		T78.9
1B.8y	Other specified iatrogenic causes		

1C	**IDIOPATHIC SHORT STATURE**		**E34.3**
1C.1	**Familial (idiopathic) short stature**		**E34.3**
1C.1a	With normal pubertal onset		
1C.1b	With delayed pubertal onset		
1C.1c	Onset of puberty not yet known		
1C.1d	Onset of puberty unknown		
1C.2	**Non-familial (idiopathic) short stature**		**E34.3**
1C.2a	With normal pubertal onset		
1C.2b	With delayed pubertal onset (constitutional delay in growth and puberty, or constitutional delay in growth and adolescence)		
1C.2c	Onset of puberty not yet known		
1C.2d	Onset of puberty unknown		

2	**TALL STATURE**		
2A	**PRIMARY GROWTH D ISORDERS**		
2A.1	**Clinically defined (dysmorphic) syndromes with sex chromosome abnormality including aneuploidy**		
2A.1a	Syndromes classified elsewhere:		
	47,XXY (Klinefelter syndrome) [primary 14A.3]		
	47,XYY syndrome [primary 14A.6]		
	Fragile X syndrome [primary 14B.14; secondary 2A.3a]		
2A.1b	47,XXX syndrome2		Q97.0
2A.1y	Other specified X and Y chromosome aneuploidy syndromes		Q97.1 Q97.8 Q98.8
2A.1z	Other syndromes, unspecified		Q97.9 Q98.9
2A.2	**Dysmorphic syndromes due to metabolic/connective tissue abnormality**		
2A.2a	Syndromes classified elsewhere:		
	Marfan syndrome (14B.20)	#154700 #154705	Q87.4
2A.2b	Marfan-like syndrome, not genetically confirmed		E72.1
2A.2c	Homocystinuria	#236250	E72.1
2A.2d	Total lipodystrophy (Berardinelli (generalised) lipodystrophy syndrome) [primary 11A.3b.5]	#236250	E88.1
2A.2y	Other specified syndromes		
2A.2z	Other syndromes, unspecified		
2A.3	**Other dysmorphic syndromes with symmetrical overgrowth**		
2A.3a	Syndromes classified elsewhere:		Q87.3
	Bannayan-Riley-Ruvalcaba syndrome (14B.4)		
	Elejalde syndrome (14B.12)		
	Fragile X syndrome (14B.14)		
	Marshall-Smith syndrome (14B.21)		
	Nevo syndrome (14B.23)		
	Simpson-Golabi-Behmel syndrome (14B.32)		
	Sotos syndrome (14B.34)		
	Weaver syndrome (14B.36)		
2A.3y	Other specified syndromes		
2A.3z	Other syndromes, unspecified		
2A.4	**Dysmorphic syndromes with partial/asymmetrical overgrowth**		**Q87.3**
2A.4a	Disorders classified elsewhere:		
	Beckwith-Wiedemann syndrome (14B.5)		
	Klippel-Trenaunay-Weber syndrome (14B.17)		
	Proteus syndrome (14B.26)		
2A.4y	Other specified syndromes		
2A.4z	Other syndromes, unspecified		

2B	**SECONDARY GROWTH D I SORDERS**		
2B.1	**Growth hormone overproduction**		**E22.0**
2B.1a	GH-producing adenoma (solitary) [primary] [secondary 6B.0]	#102200	
2B.1b	GH-producing adenoma, as part of McCune-Albright syndrome or MEN1 syndrome	#174800 +131100	Q78.1
2B.1c	GHRH excess		E22.0
2B.2	Hyperinsulinism		E15.0,E16.1
2B.2a	Primary		
2B.2a.1	Disorders classified elsewhere:		
	Transient hyperinsulinism (11B.1a) Congenital hyperinsulinism (11B.1b)		
2B.2a.9	Other disorders, unspecified		
2B.2b	Secondary		
2B.2b.1	Disorders classified elsewhere: simple obesity (5A)		
2B.2b.2	Obesity with acanthosis nigricans 8 hyperlipidaemia and sexual precocity		L83
2B.2b.9	Other disorders, unspecified		E16.1
2B.3	**Familial (isolated) glucocorticoid deficiency (ACTH insensitivity, hereditary unresponsiveness to ACTH) [primary 8A.2d]**	**#202200**	
2B.4	**Hyperthyroidism**		**E05**
2B.5	**Conditions leading to tall stature in childhood, and normal or short stature in adulthood**		
2B.5a	Diagnoses classified elsewhere:		
	Precocious puberty (3A)		
	Exogenous estrogens or androgens (3A.2d)		
	Hyperthyroidism (7B)		
	Congenital adrenal hyperplasia (8A.1)		
2B.5y	Other specified conditions		
2B.6	**Conditions leading to normal height in childhood, and tall stature in adulthood**		
2B.6a	Disorder classified elsewhere:		
	Gonadotrophin deficiency (long limbs and hypogonadism) (6A.3)		
2B.6b	Aromatase deficiency	-107910	E34.9
2B.6c	Estrogen receptor dysfunction	-133430	E34.9
2B.6y	Other specified conditions		
2C	**IDIOPATHIC (NORMAL VARIANT TALL STATURE)**		**E34.4**
2C.1	**Genetic (familial, or constitutional) tall stature**		
2C.2	**Non-familial idiopathic tall stature**		