

**WEB TABLE I** PHENOTYPE, GENOTYPE AND FINAL DIAGNOSIS OF CHILDREN WITH DISPROPORTIONATE SHORT STATURE (N=40)

<i>Phenotype</i>	<i>Genotype ( N: Number of patients )</i>	<i>Gene</i>	<i>Final diagnosis (number of cases )</i>	<i>Clue to diagnosis</i>
Large head, proximal shortening of limbs, mid face hypoplasia, brachydactyly, trident hand, progressive narrowing of interpediculate distance in lumbar spine	c. 1138 G>A, p. Gly380Arg (9 cases)	FGFR3	Achondroplasia (9)	Large head, proximal shortening of long bones
Joint immobility, short trunk, pectus carinatum, dysostosis multiplex in radiograph	c.155C>T, p.Pro52Leu (2 cases); [c.3G>A; p.1Met>Ile] [c.452 C> T ; p.Pro151Leu] (1 case)	GALNS	Morquio Syndrome (7)	Dysostosis multiplex
Coarse facies , dull look, infantile proportion (short limb dwarfism)	Not done		Congenital hypothyroidism (5)	
Short limb dwarfism, round face, flat nasal bridge, advance bone age, monkey wrench deformity of femoral head, short metacarpals, short metatarsals	c.C467T p.Ser156Ph (3)	CANT1	Desbuquois dysplasia (3)	Monkey wrench deformity of femoral head, advanced carpal age
Ovoid vertebrae, metaphyseal flaring, epiphyseal flaring, joint laxity, onset of disease after 2 years of age, Normal shape of body unlike achondroplasia.	c.1554C>G ; p.Asp518Glu (1)	COMP;	Pseudoachondroplasia (2)	Onset of disease after 2 years of age
Short trunk dwarfism, block/hemivertebrae (thoracic), multiple rib defects (fused) , absence of finger anomalies	Not done		Spondylocostal dysostosis IV(1)	Involvement of thoracic spine and ribs (asymmetrical)
Short trunk dwarfism, block / hemivertebrae (thoracic ) , no intrinsic rib defects, absence of finger anomalies, ribs are fused symmetrically to the spine posteriorly (crab like chest deformity)	Not done		Spondylothoracic dysplasia (1)	Involvement of spine and ribs (symmetrical)> crab like chest deformity
Short trunk dwarfism , microcephaly, severe mental retardation, wavy iliac crest (pathognomic)	c.1923del,p.(Tyr642 Metfs*78)(1)	DYM, Dymeclin	Dyggve Mechoir Clausen disease (1)	Wavy iliac crest and mental retardation
Small thorax, rhizomelia, proximally placed thumb, talipes equinovarus, bifid distal humerus	Not done		Atelosteogenesis II (1)	Bifid distal humerus
Short trunk dwarfism, block / hemivertebrae (thoracic), multiple rib defects (fused) , absence of finger anomalies	Not done		Spondylocostal dysostosis (1)	Involvement of thoracic spine and ribs (asymmetrical)

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<i>Phenotype</i>	<i>Genotype ( N: Number of patients )</i>	<i>Gene</i>	<i>Final diagnosis (number of cases )</i>	<i>Clue to diagnosis</i>
Short limb dwarfism, fine hair, brittle hair, metaphyseal involvement	g.69dupG,r.69dupG(1)	RMRP	Cartilage Hair hypoplasia (1)	Fine hair, only metaphyseal flaring in long bones
Short limb dwarfism, narrow thorax, long coccyx, coccygeal tail, delayed bone age	Not done		Metatropic dysplasia (1)	Presence of coccygeal tail
Short limb dwarfism (acral and mesomelia) , loose and redundant skin on fingers, metaphyseal flaring, ovoid vertebrae	Not done		Acromesomelic dysplasia (1)	Acral and mesomelic parts of long bones are severely affected.
Short limb and short trunk, metaphyseal flaring, ovoid vertebrae	Not done		Unclassified (6)	Needs whole exome/ genome sequencing.