

Table 1. Patients with positive genetic results

	Gene	Variant	Zygoty	Variant classification	Inheritance	Clinical features*	Clinical impact	
							Change in management	Genetic counselling for family planning
Case 2	<i>KIF1A</i>	c.757G>A (p.Glu253Lys)	Heterozygous	Pathogenic	De novo	Developmental delay , intellectual disability, peripheral neuropathy, microcephaly, distal arthrogryposis	<input checked="" type="checkbox"/> Allowed holistic management of the child with surveillance for complications such as eye review for optic atrophy and visual impairment.	<input checked="" type="checkbox"/>
Case 5	<i>IFT122</i>	c.1980C>A (p.Tyr660Ter)	Heterozygous	Pathogenic	Paternal	Cranioectodermal dysplasia: skeletal dysplasia , ectodermal defects, joint laxity	<input type="checkbox"/> Not applicable	<input checked="" type="checkbox"/>
		c.694G>A (p.Gly232Ser)	Heterozygous	Likely pathogenic	Maternal			
Case 6	<i>SAMD9</i>	c.3406G>C (p.Glu1136Gln)	Heterozygous	Likely pathogenic	De novo	MIRAGE (Myelodysplasia, infection, restriction of growth, adrenal hypoplasia, genital phenotypes and enteropathy) syndrome: developmental delay, adrenal insufficiency, genital underdevelopment, thrombocytopaenia and/or anaemia, severe invasive infections and immunologic abnormalities.	<input checked="" type="checkbox"/> Allowed holistic and targeted management, with surveillance for other complications, including haematological malignancy and immunodeficiency.	<input checked="" type="checkbox"/>
Case 8	<i>FGFR3</i>	c.1620C>A (p.Asn540Lys)	Heterozygous	Pathogenic	De novo	Hypochondroplasia: short stature, lumbar lordosis , short and broad bones, caudad narrowing of the interpediculate distance of the lumbar spine.	<input checked="" type="checkbox"/> Surveillance for skeletal complications of hypochondroplasia.	<input checked="" type="checkbox"/>
Case 10	<i>SHOC2</i>	c.4A>G (p.Ser2Gly)	Heterozygous	Pathogenic	De novo	Noonan-like syndrome with loose anagen hair: short stature, cognitive deficits, cardiac defects , ectodermal abnormalities.	<input checked="" type="checkbox"/> Allowed holistic and targeted management, with surveillance for other complications such as short stature and need for regular growth monitoring.	<input checked="" type="checkbox"/>

* Clinical features in bold are those seen in our patients

 present absent