Table S4: Clinical presentation of patients associated with *IFT81* gene in literature

Literature	Allele 1	Allele 2	Retinal Dystrophy	History of Speech Delay	Additional Findings	Last Evaluation	Interpretation
This study (P7)	Exon 19 c.1969C>T (p.Gln657Ter)	Exon 19 c.1969C>T (p.Gln657Ter)	+ (Age of onset 7 years)	+	-	26 years	*IFT81*-mediated nonsyndromic IRD
Perea-Romero et al. PMID: 34448047	Exon 19 c.1969C>T (p.Gln657Ter)	Exon 19 c.1969C>T (p.Gln657Ter)	+ (Age of onset NA)	NA	Mixed HI, DI, HH, joint rheumatism, osteopenia, spinal osteoarthritis, oligospermia, abnormality of higher mental functions	NA	There are visual and skeletal findings association with *IFT81* in the same patient Likely pathogenic
Ashraf et al. PMID: 32783357	Exon 19 c.1934_1937delinsGAAG (p.Leu645Ter)	Intron 14 c.1557+3_1557+6del	-	+	Short-rib polydactyly syndrome, rectal atresia, situs inversus, micropenis	29 months	Unique features
Perrault et al. PMID: 26275418	Intron 11 c.1188+1G>A	Intron 11 c.1188+1G>A	-	+	Postaxial polydactyly, moderate ID, NPHP	5 years	The patient may develop retinal dystrophy later in life Hypomorphic allele
	Exon 19 c.2015_2019del (p.Asp672AlafsTer15)	Exon 19 c.2015_2019del (p.Asp672AlafsTer15)	+ (Age of onset 4 years)	+	Mild ID, diffuse cerebellar atrophy, night enuresis, polyuria and polydipsia	11 years	Hypomorphic allele
Duran et al. PMID: 27666822	Exon 2 c.87G>C (p.Leu29Phe)	Exon 14 c.1534C>T p.(Arg512Ter)	NA	NA	Asphyxiating thoracic dysplasia	Died at 19 months of age	Mutations in *IFT81* destabilize the anterograde transport complex, and this is considered to be the mechanism in skeletal ciliopathies
	Exon 9 c.785T>G (p.Leu262Ter)	Exon 12 c.1303_1305delCTT (p.Leu435del)	NA	NA	Short-rib polydactyly syndrome	Died a few minutes after birth	
Dharmat et al. PMID: 28460050	Exon 12 c.1213C>T (p.Arg405Ter)	Exon 18 c.1841T>C (p.Leu614Pro)	+ (Age of onset 12 years)	-	-	22 years	*IFT81* is a candidate gene for nonsyndromic IRD, c.1841T>C is likely to be a hypomorphic allele

Abbrevations: NA: non available, HI: hearing impairment, DI: diabetes insipidus, HH: hypothalamic hypothyroidism, ID: intellectual disability, NPHP: nephronophthisis,