

Table S1. *MKS1* mutations in individuals with Meckel syndrome

Subject	Origin	cDNA change NM_017777.3	Protein change	EGA	MTS	OE	Ret	Col	Kid	Liver	PD	Other
Frank '07: 850	Turkish	c.262-37_179del c.262-37_179del	p.F88_E139del p.F88_E139del	ND	ND	+	ND	ND	+	+	+	DWM, hydrocephalus, cleft lip
Frank '07: 937	Turkish	c.1407+2delT c.1407+2delT	Splice Splice	ND	ND	+	ND	ND	+	+	+	
Frank '07: 951	Kuwaiti	c.515+1G>A c.515+1G>A	Splice Splice	ND	ND	+	ND	ND	+	+	+	
Frank '07: 943	German	c.1408-35del29 c.417G>A	p.G471Lfs*92 p.F88_E139del ²	ND	ND	+	ND	ND	+	+	+	
Khaddour '07: 20	French	c.417G>A c.424C>T	p.F88_E139del ² p.Q142*	15	ND	+	ND	ND	+	+	+	Cleft palate, situs inversus, skeletal dysplasia
Khaddour '07: 362	French	c.958G>A ³ c.1408-35del29	p.V320I(splice) p.G471Lfs*92	26	VA	+	ND	ND	+	+	+	Cleft palate, arhinencephaly, ACC, pancreatic cysts
Khaddour '07: 433	French	c.184_190del7 c.1490G>A ³	p.T61Vfs*14 p.R497K(splice)	ND	ND	+	ND	ND	+	ND	+	
Khaddour '07: 434	French	c.184_190del7 c.1490G>A ³	p.T61Vfs*14 p.R497K(splice)	14	ND	+	ND	ND	+	ND	+	
Khaddour '07: 522	Palestinian	c.1048C>G c.1048C>G	p.Q350* p.Q350*	Term	ND	+	ND	ND	+	ND	+	
Khaddour '07: 523	Palestinian	c.1048C>G c.1048C>G	p.Q350* p.Q350*	Term	ND	+	ND	ND	+	ND	+	Ulnar bowing
Khaddour '07: 532	Palestinian	c.1048C>G c.1048C>G	p.Q350* p.Q350*	Term	ND	+	ND	ND	+	+	+	
Khaddour '07: 533	Palestinian	c.1048C>G c.1048C>G	p.Q350* p.Q350*	Term	ND	+	ND	ND	+	ND	+	Femoral bowing
Khaddour '07: 534	Palestinian	c.1048C>G c.1048C>G	p.Q350* p.Q350*	Term	ND	+	ND	ND	+	ND	+	
Khaddour '07: 562	French	c.472C>T c.1408-35del29	p.R158* p.G471Lfs*92	13	ND	ND	ND	ND	+	+	+	Cleft palate, IUGR, micromelia, hygroma
Khaddour '07: 106	English	c.1408-35del29 c.1408-35del29	p.G471Lfs*92 p.G471Lfs*92	19	ND	+	ND	ND	+	+	+	Epididymal cysts
Khaddour '07: 102	Pakistani	c.1448_1451dupCAGG c.1448_1451dupCAGG	p.T485Rfs*107 p.T485Rfs*107	18	ND	+	ND	ND	+	+	+	Cleft palate, situs inversus, skeletal dysplasia
Auber '07: 1	German	c.1408-35del29 c.1408-35del29	p.G471Lfs*92 p.G471Lfs*92	21	ND	+	ND	+	+	+	+	Cleft palate, campomelia
Auber '07: 2	German	c.1408-35del29 c.1408-35del29	p.G471Lfs*92 p.G471Lfs*92	31	ND	+	ND	+	+	+	+	Epididymal cysts, polysplenia,

												hypoplastic left heart
Auber '07: 3	German	c.1408-35del29 c.1408-35del29	p.G471Lfs*92 p.G471Lfs*92	18	ND	+	ND	+	+	+	+	Campomelia
Auber '07: 4	German	c.1408-35del29 c.1408-35del29	p.G471Lfs*92 p.G471Lfs*92	19	ND	FMC	ND	-	+	+	+	Campomelia, epididymal cysts, partial agenesis of the corpus callosum
Auber '07: 5	German	c.1408-35del29 c.1408-35del29	p.G471Lfs*92 p.G471Lfs*92	19	ND	+	ND	+	+	+	+	Cleft palate, campomelia, epididymal cysts, Robin sequence, holoprosencephaly
Auber '07: 6	German	c.1408-35del29 c.1408-35del29	p.G471Lfs*92 p.G471Lfs*92	18	ND	+	ND	+	+	+	+	Campomelia, epididymal cysts, ambiguous genitalia
Auber '07: 7	German	c.1408-35del29 c.1408-35del29	p.G471Lfs*92 p.G471Lfs*92	22	ND	+	ND	ND	+	+	+	Cleft palate, campomelia, epididymal cysts,
Auber '07: 8	German	c.1408-35del29 c.1408-35del29	p.G471Lfs*92 p.G471Lfs*92	24	ND	+	ND	ND	+	+	+	Cleft palate, epididymal cysts, horseshoe kidney
Kyttälä '06: F1	German	c.1408-35del29 c.1408-35del29	p.G471Lfs*92 p.G471Lfs*92	ND	ND	ND	ND	ND	ND	ND	ND	
Kyttälä '06: F2	Mixed European/ Portuguese	c.1408-35del29 c.1408-35del29	p.G471Lfs*92 p.G471Lfs*92	ND	ND	ND	ND	ND	ND	ND	ND	
Kyttälä '06: F3	U.S.	c.1408-35del29 c.1408-35del29	p.G471Lfs*92 p.G471Lfs*92	ND	ND	ND	ND	ND	ND	ND	ND	
Kyttälä '06: F4	German	c.50insCCGGG ⁴ c.80+2T>C ⁵	p.R17Rfs*163 ⁴ Splice	ND	ND	ND	ND	ND	ND	ND	ND	
Consugar '07: M338	AA/Caucasian	c.1099+1G>A c.1408-35del29	Splice p.G471Lfs*92	16	ND	+	ND	ND	+	ND	+	
Consugar '07: M340	German	c.1408-35del29 c.417G>A	p.G471Lfs*92 p.F88_E139del	15	ND	+	ND	ND	+	ND	+	
Consugar '07: M380	Mixed European/ Native American	c.1408-35del29 c.1408-35del29	p.G471Lfs*92 p.G471Lfs*92	19	ND	+	ND	ND	+	ND	+	
Consugar '07: M383	Mixed European	c.1408-35del29 c.1408-35del29	p.G471Lfs*92 p.G471Lfs*92	11	ND	+	ND	ND	*	ND	+	

Consugar '07: 55875	Dutch	c.417G>A c.1408-35del29	p.F88_E139del p.G471Lfs*92	16	ND	+	ND	ND	+	+	+	
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¹PolyPhen-2 scores (HumDiv/HumVar)

²Based on RT-PCR data in Consugar et al. 2007

³Last base pair in exon, predicted to affect splicing

⁴designated 50insCCGGG; P17fsX163 in Kyttälä et al 2006

⁵designated IVS1+2T->C in Kyttälä et al 2006

EGA: estimated gestational age; FMC: Foramen Magnum Cephalocele; Term: baby was born and died several hours/days later; ND: not documented; ?: unknown; MTS: molar tooth sign; OE: occipital encephalocele; Ret: retinal dystrophy; Col: coloboma; Kid: kidney disease; Liver: liver fibrosis; PD: polydactyly; DWM: Dandy-Walker malformation; ACC: agenesis of the corpus callosum; VA: vermis aplasia; AA: African American; IUGR: intrauterine growth restriction; U.S.: United State