

Table S2. Detailed clinical summary of the affected fetuses of families 22 and 23 with variants in *KIF21A* (NM\_001173464.2)

Family Genotype	Family 22 c.1346T>A, (p.Leu449*) homozygous				Family 23 c.2371del, (p.Arg791Glufs*8) homozygous				
	22:II.1 male		22:II.2 female		22:II.4 male		23:II.1 male		23:II.2 female
	prenatal ultrasound (WGA 22+2)	autopsy (WGA 23+5)	prenatal ultrasound (WGA 23+3)	autopsy (WGA 25+1)	prenatal ultrasound (WGA 19+0)	prenatal ultrasound (WGA 25+5)	autopsy (WGA 30+3)	prenatal ultrasound (WGA 21+0)	
Investigation	Yes	–	Yes	–	Yes	–	–	Yes	
RFM	–	–	Yes	–	–	–	–	–	
IUGR	–	–	Yes	–	–	–	–	–	
Polyhydramnios	Yes	–	Yes	–	–	–	Yes	–	
Cerebral ventriculomegaly	–	–	–	–	–	–	Yes	ND (autolysis)	
Brachycephaly	–	–	–	–	–	–	Yes	ND (autolysis)	
Scalp edema	Yes	–	Yes	–	–	–	Yes	Yes (with protruding skull bones)	
Neck edema	–	–	–	–	Yes	–	Yes	–	
Hypertelorism	–	Yes	–	–	–	–	–	–	
Low-set ears	–	Yes	Yes	ND	–	–	–	ND	
Flat broad nose	–	Yes	–	–	ND	–	–	–	
Prenasal edema	–	–	–	–	ND	–	Yes	ND (autolysis)	
Cleft-palate of the hard and soft palate	–	Yes (hard and soft palate)	–	Yes (median cleft of the soft palate)	–	–	ND	–	
Microglossia	–	–	–	Yes	–	–	–	–	
Micrognathia	–	–	Yes	Yes	Yes	–	Yes	Yes	
Retrognathia	Yes	Yes	Yes	Yes	–	–	Yes	Yes	
High-arched palate	–	–	–	Yes	–	–	–	–	
Thoracic / Pulmonary hypoplasia	Yes	Yes	Yes	Yes	Yes	–	Yes	Yes	
Hydrothorax	–	–	Yes	–	–	–	–	–	
Cardial abnormalities	–	–	–	–	–	–	Yes (dextrocardia)	Yes (dextrocardia)	
Gastrointestinal abnormalities	Yes (diaphragmatic protrusion, missing stomach filling)	ND (autolysis)	Yes (diaphragmatic protrusion, missing stomach filling, ascites)	Jejunal perforation	ND	–	–	Yes (missing stomach filling)	
Urogenital abnormalities	–	–	–	Dilated tortuous ureter (bilateral)	–	–	–	–	
Abnormalities of joints / spine / muscles:									
Anomalies of the spine	Yes (thoracic kyphoscoliosis)	Yes (thoracic kyphoscoliosis)	–	Yes (thoracic scoliosis, additional cervical rib (origin: sixth cervical vertebrae))	ND	–	–	–	
Straight ribs	–	Yes	–	Yes	–	–	–	–	
Narrow long tubular bones	–	Yes	–	Yes	–	–	–	–	
Internally rotated shoulders	–	Yes (bilateral)	–	Yes (bilateral)	–	–	–	–	
Elbow fixed in flexion	Yes	–	Yes	–	–	–	–	–	
Elbow fixed in extension	–	Yes	–	Yes	–	–	–	–	
Wrist fixed in flexion	Yes	Yes	–	–	–	–	Yes (bilateral)	Yes (bilateral)	
Clinodactyly	–	–	–	–	–	–	Yes (bilateral)	Yes (DII, bilateral)	
Clenched hands with crossing fingers	Yes	Yes (with bilateral infolded thumb; left: DII over DI, DIII-DV fixed in middle joint; right: DIII-DV over DI, DII fixed in middle joint)	Yes (bilateral)	Yes (with bilateral infolded thumb; left: DII-DIII over DI; right: DII-DV over DI)	–	–	Yes (bilateral)	Yes (with infolded thumb, bilateral)	
Femoral anteversion	–	–	–	Yes (bilateral)	–	–	–	–	
Knee joint fixed in extension	Yes	Yes (bilateral)	Yes (bilateral)	Yes (bilateral)	Yes (bilateral)	–	–	Yes (unilateral)	
Knee joint fixed in flexion	–	–	–	–	–	–	–	Yes (unilateral)	
Genu recurvatum	–	–	–	Yes (bilateral)	–	–	–	–	
Pes equinovarus	Yes (bilateral)	Yes (bilateral)	Yes (bilateral)	Yes (bilateral)	Yes (bilateral)	Yes (bilateral)	Yes (bilateral)	Yes (bilateral)	
Rocker bottom foot	–	–	–	–	–	–	Yes (bilateral)	–	
Hypotrophic muscles of the lower legs	Yes	–	–	–	–	–	–	–	

ND no data, IUGR intrauterine growth restriction, RFM reduced fetal movements, WGA weeks of gestational age