Table S2. Detailed clinical summary of the affected fetuses of families 22 and 23 with variants in *KIF21A* (NM_001173464.2)

Family	Family 22					Family 23		
Genotype	c.1346T>A, (p.Leu449*) homozygous					c.2371del, (p.Arg791Glufs*8) homozygous		
Patient ID	22:II.1		22:II.2		22:II.4	23:II.1		23:II.2
Sex	male		female		male	male		female
Investigation	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)
RFM	Yes	–	Yes	–	Yes	Yes	–	Yes
IUGR	–	–	Yes	–	–	–	–	–
Polyhydramnios	Yes	–	Yes	–	–	Yes	–	Yes
Cerebral venticulomegaly	–	–	–	–	–	Yes	ND (autolysis)	–
Brachycephaly	–	–	–	–	–	Yes	ND (autolysis)	–
Scalp edema	Yes	–	Yes	–	–	Yes	Yes (with protruding skull bones)	Yes
Neck edema	–	–	–	–	Yes	Yes	–	–
Hypertelorism	–	Yes	–	–	–	–	–	–
Low-set ears	–	Yes	Yes	ND	–	–	–	ND
Flat broad nose	–	Yes	–	–	ND	–	–	–
Prenasal edema	–	–	–	–	ND	Yes	ND (autolysis)	Yes
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	Yes
Retrognathia	Yes	Yes	Yes	Yes	–	Yes	Yes	Yes
High-arched palate	–	–	–	Yes	–	–	–	–
Thoracic / Pulmonary hypoplasia	Yes	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)	Yes (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