Table S1. Clinical details of investigated individuals without molecular cause identified

Patient ID #	Family #	Gender / age at inclusion termination of pregnancy	Affected gene	Prenatal abnormalities	Joint contractures (prenatal or postnatal)	Dysmorphic features	Neurological abnormalities	Other features
F16:II.1	16	m / 3 mo	no causal variant found	RFM, genu valgum	Elbows, wrists, knees, talipes equinovarus (bilateral)	Sandal gap (bilateral)	–	–
F17:II.1	17	m / TOP (35 WGA)	no causal variant found	RFM	Elbows, wrists, knees, talipes equinovarus (bilateral)	ND (autopsy not performed)	–	–
F18:II.1	18	f / 3 y	no causal variant found	RFM, polyhydramnios	Elbows, hips, knees, talipes equinovarus (bilateral)	Naevus flammeus, joint dimples	Global developmental delay, choroid plexus cyst, muscle weakness	Hip luxation (bilateral), deformed ribs, scoliosis
F19:II.1	19	f / TOP (22 WGA)	no causal variant found	RFM, vitium cordis, shortened trunk	Fingers, wrists, knees, crossed fingers, pointed feet	ND (autopsy not performed)	Spina bifida, corpus callosum agenesia, severe spinal deformation	–
F20:II.1	20	m / TOP (19 WGA)	no causal variant found	RFM, IUGR, hydrothorax, singular umbilical cord	Elbows, knees, talipes equinovarus (bilateral)	Retrognathia, syndactyly left hand (3th-5th digit), prominent nasal root	–	Anal atresia, pulmonary hypoplasia, hydrothorax, pericardial effusion, malrotation of the bowel, spherical pancreas, horseshoe kidneys, dilated heart atria, agenesis of the left umbilical artery, scoliosis, absence of os frontale (bilateral)
F21:II.1	21	f / 8 y	no causal variant found	RFM, oligohydramnios, short long tubular bones	Elbows, knees, hips, talipes equinovarus (bilateral)	–	Cognitive impairment	Hip luxation

List of genetic variants and clinical details of the cohort sorted by disease-causal genetic defects in known FA-associated genes, cases with variants of unknown significance, unsolved patients, and patients carrying bi-allelic variants in the candidate gene *KIF21A*. Provided information include patient ID, family ID, gender, age at inclusion or death in months and years or termination of pregnancy in weeks of gestational age. Age 0 denotes children dying < 1 month after birth. Additionally, provided data include the affected gene, RefSeq ID, DNA change, AA change, variant classification (ACMG scores), gene-associated OMIM-phenotypes, mode of inheritance, and zygosity. Furthermore, it is indicated whether variants have already been published independently (with corresponding PubMed ID) or not. The variant nomenclature is in accordance with Human Genome Variation Society (HGVS) standards.

ǂ death, *AA* amino acid, *ACMG* American College of Medical Genetics and Genomics, *AD* autosomal dominant, *AR* autosomal recessive, *f* female, *IUGR* intrauterine growth restriction, *m* male, *mo* month, *ND* no data, *NIV* non-invasive ventilation, *OMIM* Online Mendelian Inheritance in Man, *PMID* PubMed ID, *RFM* reduced fetal movements, *RI* respiratory insufficiency, *TOP* termination of pregnancy, *VUS* variant of uncertain significance in the ACMG classification system, *WGA* weeks of gestational age, *y* years.

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