Case	Prenatal phenotype (HPO term)	Pregnancy outcome (GA)	Postnatal/postmortem phenotype (HPO term)	pES analysis CNV or Gene (OMIM) Variation cDNA & protein (SNV) Inheritance Related phenotype (OMIM)	Reanalysis CNV or Gene (OMIM) Variation cDNA & protein (SNV) Inheritance Related phenotype (OMIM)	Additional investigations	Conclusion reanalysis
Patient 4	Fetal ascites HP:0001791 Pleural effusion HP:0002202 Polyhydramnios HP:0001561 Hydrops fetalis HP:0001789 Congenital diaphragmatic hernia HP:0000776	COP Live birth (36)	No (neonatal death)	del1q21.1 inherited (VUS) *(145414780–145826931)x1* Maternally inherited	No relevant variant	No	Negative Del1q21.1 excluded
Patient 14	Tetralogy of Fallot HP:0001636 Dandy-Walker malformation HP:0001305 Hypoplasia of the brainstem HP:0007362	TOP (32)	Rhizomelia HP:0008905 Delayed skeletal maturation HP:0002750 Prominent sternum HP:0000884 Hypertelorism HP:0000316 Abnormal nasal morphology HP:0005105	*CUX1* NM_001913: c.1671C>A, p.Tyr557* AD, maternally inherited Global developmental delay with or without impaired intellectual development (MIM#618330)	No relevant variant	No	Negative *CUX1* variant excluded
Patient 139	Hemivertebrae HP:0002937 Talipes equinovarus HP:0001762 Lateral ventricular asymmetry HP:0100960 Dysostosis multiplex HP:0000943	TOP (36)	No postmortem examination	dupXq28 (148564275–148798438)x3 Maternally inherited	dupXq28 (148564275–148798438)x3 Maternally inherited VUS *SGSH (*NM_000199:c.505C>G, p.Arg169Gly) identified by reanalysis (1 hit)	Biochemical tests	Negative CNV downgraded following biochemical test
Patient 73	Abdominal situs inversus HP:0003363 Previous pregnancy: TOP for heterotaxia and complex heart defect	COP Live birth (41)	No additional finding (M24)	*DNAH11* (606582) NM_001277115: c.1848+1G>T, p.? NM_001277115: c.10472G>A, p.Arg3491His AR, Compound heterozygous Ciliary dyskinesia primary 7 with or without situs inversus (#MIM611884)	*DNAH11* (606582) NM_001277115: c.1848+1G>T, p.? NM_001277115: c.10472G>A, p.Arg3491His AR, Compound heterozygous Ciliary dyskinesia primary 7 with or without situs inversus (#MIM611884)	Terminated fetus with biallelic *DNAH11* variant Functional study	VUS
Patient 87	Cerebellar vermis hypoplasia HP:0001320 Abnormal cortical gyration HP:0002536	COP Live birth (26)	M12 Motor delay HP:0001270	*DLL1* (606582) *NM_005618:c.553G>T,* p.Gly185* AD, maternally inherited Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizure (MIM#618709)	*DLL1* (606582) *NM_005618:c.553G>T,* p.Gly185* AD, inherited from mother Neurodevelopmental disorder with nonspecific brain abnormalities and with or without seizure (MIM#618709)	Familial segregation	VUS
Patient 94	Agenesis of corpus callosum HP:0001274 Enlarged fetal cisterna magna HP:0011427 Bicuspid aortic valve HP:0001647 Lissencephaly HP:0001339	TOP (30)	Macrocephaly HP:0000256 Hypertelorism HP:0000316 Microretrognathia HP:0000308 Low-set ears HP:0000369	*KMT2E* (608444) NM_018682: c.5219delA, p.His1740fs AD, maternally inherited Homozygous with isodisomy 7q O'Donnell Luria Rodan syndrome (MIM#618512)	*KMT2E* (608444) NM_018682: c.5219delA, p.His1740fs AD, maternally inherited Homozygous with isodisomy 7q O'Donnell Luria Rodan syndrome (MIM#618512)	Methylation signature	VUS
Patient 123	Tetralogy of Fallot HP:0001636 Abnormal male external genitalia morphology HP:0000032	TOP (26)	Micropenis HP:0000054 Low-set ears HP:0000369 Retrognathia HP:0000278 Short philtrum HP:0000322 Hypertelorism HP:0000316 Long eyelashes HP:0000527 Sloping forehead HP:0000340 Single umbilical artery HP:0001195	*KAT7* (609880) NM_007067:c.1480A>G, p.Ser494Gly *de novo* Not related to human disease	*KAT7* (609880) NM_007067:c.1480A>G, p.Ser494Gly *de novo* Not related to human disease	Replication / datasharing	VUS

			Microcornea HP:0000482				
Patient 145	Agenesis of corpus callosum HP:0001274	TOP (33)	No (postmortem examination not accepted)	*del6p21.32* (32940674–32947911)x1 *De novo* *BRD2* gene	*del6p21.32* (32940674–32947911)x1 *De novo* *BRD2* gene	Replication / datasharing	VUS
Patient 22	Postaxial foot polydactyly HP:0001830	COP Live birth (39)	No additional finding (M24)	*Negative*	*GLI1* NM_005269:c.535-3A>G Paternally inherited	*RT-PCR*	VUS
Patient 104	Ureteral duplication HP:0000073 Talipes equinovarus HP:0001762 Single umbilical artery HP:0001195	COP Live birth (38)	Patent ductus arteriosus (HP:0001643) Retrognatia (HP:0000278) Feeding difficulties (HP:0011968) Speech delay (HP:0000750) Epicanthus (HP:0000286) Tented upper lip vermilion (HP:0010804) Broad eyebrow (HP:0011229)	*Negative*	*SYNE1* NM_182961: c.20779G>C, p.Glu6927Gln NM_182961: c.12820C>A, p.His4274Asn AR, Compound heterozygous Arthrogryposis multiplex congenita 3, myogenic type 618484	Replication / datasharing	VUS
Patient 18	Increased nuchal translucency HP:0010880 Duodenal atresia HP:0002247 Tetralogy of Fallot HP:0001636 Absent gallbladder HP:0011467 Micropenis HP:0000054	TOP (32)	Annular pancreas HP:0001734 Polysplenia HP:0001748 Intestinal malrotation HP:0002566 Left Isomerism HP:0031854 Persistent left superior vena cava HP:0005301 Hypoplasia of the gallbladder HP:0005233	*Negative*	*MMP21* NM_147191:c.1203G>A p.Trp401* Heterotaxy visceral 7 616749	Genome sequencing	Negative No second variant in sr-GS ddRT-PCR excluded biallelic loss of function