

Patient	Related parents	GA at first ultrasound findings (WG)	Prenatal findings	pES result	Pregnancy outcome (WG)	Additional postnatal / postmortem data	Follow-up (Age in month)
Patient 4	No	22	Fetal ascites HP:0001791 Pleural effusion HP:0002202 Polyhydramnios HP:0001561 Hydrops fetalis HP:0001789 Congenital diaphragmatic hernia HP:0000776	del1q21.1	COP Live birth (36)	No	Neonatal death at Day 1
Patient 8	No	20	Microcephaly HP:0000252 Short corpus callosum HP:0200012 Abnormal septum pellucidum morphology HP:0007375 Cerebellar cyst HP:0002350 Abnormal cortical gyration HP:0002536	negative	TOP (35)	Polymicrogyria HP:0002126 Subependymal cysts HP:0002416 Microretrognathia HP:0000308 Intracerebral periventricular calcifications HP:0007229 Cerebral calcification HP:0002514	NA
Patient 9	No	20	Intrauterine growth retardation HP:0001511 Echogenic fetal bowel HP:0010943 Short long bone HP:0003026 Micropenis HP:0000054	negative	COP Live birth (33)	Poor suck HP:0002033	M35 Sucking-swallowing disorders
Patient 11	No	21	Large for gestational age HP:0001520 Agenesis of corpus callosum HP:0001274	negative	COP Live birth (40)	No	M35 Hypertrichosis HP:0000998 Global developmental delay HP:0001263 Epicanthus HP:0000286 Generalized hypotonia HP:0001290 Low frustration tolerance HP:0000744 Short attention span HP:0000736
Patient 14	No	15	Tetralogy of Fallot HP:0001636 Dandy-Walker malformation HP:0001305	VUS <i>CUX1</i>	TOP (32)	Rhizomelia HP:0008905 Delayed skeletal maturation HP:0002750 Prominent	NA

			Hypoplasia of the brainstem HP:0007362			sternum HP:0000884 Hypertelorism HP:0000316 Abnormal nasal morphology HP:0005105	
Patient 18	No	11	Increased nuchal translucency HP:0010880 Duodenal atresia HP:0002247 Tetralogy of Fallot HP:0001636 Absent gallbladder HP:0011467 Micropenis HP:0000054	negative	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	NA
Patient 21	No	21	Pierre-Robin sequence HP:0000201 Adactyly HP:0009776	negative	TOP (33)	No	NA
Patient 22	No	16	Postaxial foot polydactyly HP:0001830	negative	COP Live birth (39)	No	M24 No
Patient 23	No	20	Oligohydramnios HP:0001562 Intrauterine growth retardation HP:0001511 Hypoplasia of the corpus callosum HP:0002079	negative	TOP (31)	CMV infection Confirmed in amniotic fluid and fetal tissues	NA
Patient 28	No	20	Intrauterine growth retardation HP:0001511 Duodenal atresia HP:0002247 Tetralogy of Fallot HP:0001636	negative	IUFD (33)	Fetal pleural effusion HP:0025676 Abnormal lung lobation HP:0002101	NA
Patient 32	No	20	Fetal bowel dilatation HP:4000140 Anal atresia HP:0002023 Postaxial foot polydactyly HP:0001830	negative	TOP (30)	Bifid uvula HP:0000193 Rectovaginal fistula HP:0000143 Left aortic arch with retroesophageal right subclavian artery HP:0011595 Aplasia/Hypoplasia of the gallbladder HP:0011466 Aplasia/Hypoplasia of the fifth metatarsal bone HP:0040033 Hydronephrosis HP:0000126	NA

Patient 33	No	21	Cleft lip HP:0410030 Ureteral duplication HP:0000073	negative	COP Live birth (41)	No	Clinical follow-up not requested by parents
Patient 39	No	20	Increased nuchal translucency HP:0010880 Intrauterine growth retardation HP:0001511 Lateral ventricular asymmetry HP:0100960 Pericardial effusion HP:0001698 Hypospadias HP:0000047	negative	COP Live birth (38)	CMV infection Confirmed in amniotic fluid and fetal tissues	NA
Patient 43	No	21	Oligohydramnios HP:0001562 Hyperechogenic kidneys HP:0004719 Abnormal calvaria morphology HP:0002683	negative	COP Live birth (31)	No	M25 No additional finding
Patient 45	No	30	Polyhydramnios HP:0001561 Hydrops fetalis HP:0001789 Chylothorax HP:0010310	negative	COP Live birth (37)	Intrauterine growth retardation HP:0001511 Poor suck HP:0002033	M28 Tracheomalacia HP:0002779 Generalized hypotonia HP:0001290 Neurodevelopmental delay HP:0012758 Plagiocephaly HP:0001357 Flat face HP:0012368 Single transverse palmar crease HP:0000954 High palate HP:0000218 Low frustration tolerance HP:0000744
Patient 48	No	20	Cleft palate HP:0000175 Short thumb HP:0009778 Ventricular septal defect HP:0001629 Unilateral renal agenesis HP:0000122	negative	TOP (30)	Supernumerary ribs HP:0005815 Hemivertebrae HP:0002937	NA
Patient 50	No	20	Intrauterine growth retardation HP:0001511 Abnormal heart morphology HP:0001627	negative	IUFD (29)	No	NA

			Hypoplasia of the semicircular canal HP:0011382				
Patient 52	No	24	Agenesis of corpus callosum HP:0001274	negative	TOP (34)	Atrial septal defect HP:0001631 Hypoplasia of the thymus HP:0000778	NA
Patient 54	No	30	Agenesis of corpus callosum HP:0001274	negative	COP Live birth (38)	No	M28 Bulbous nose HP:0000414 Low frustration tolerance HP:0000744 Hypoplasia of the frontal lobes HP:0007333 Sleep abnormality HP:0002360 HP:0012433 Abnormal social behavior
Patient 55	No	17	Intrauterine growth retardation HP:0001511 Partial agenesis of the corpus callosum HP:0001338	negative	COP Live birth (37)	No	Clinical follow-up not requested by parents
Patient 57	No	24	Craniosynostosis HP:0001363 Persistent left superior vena cava HP:0005301 Single umbilical artery HP:0001195	negative	COP Live birth (39)	Intrauterine growth retardation HP:0001511 Scaphocephaly HP:0030799 Hypospadias HP:0000047 Ventricular septal defect HP:0001629 Strabismus HP:0000486 Axial hypotonia HP:0008936	M20 No additional finding
Patient 61	No	16	Agenesis of corpus callosum HP:0001274 ? Inferior cerebellar vermis hypoplasia HP:0007068	negative	COP Live birth (38)	Poor suck HP:0002033 Sucking-swallowing disorders	M23 Median cleft palate HP:0009099 Clinodactyly HP:0030084 Hypertelorism HP:0000316 Frontal bossing HP:0002007 Inferior cerebellar vermis hypoplasia HP:0007068 Aplasia of the olfactory bulb HP:0032466

							Neurodevelopmental delay HP:0012758
Patient 65	No	20	Agenesis of corpus callosum HP:0001274	negative	COP Live birth (41)	No	Loss of follow-up
Patient 63	No	10	Arthrogryposis multiplex congenita HP:0002804 Gastroschisis HP:0001543	negative	COP Live birth (34)	No	Neonatal death
Patient 67	No	10	Large for gestational age HP:0001520 Agenesis of corpus callosum HP:0001274	negative	COP Live birth (41)	No	M20 Cryptorchidism HP:0000028
Patient 73	No	18	Abdominal situs inversus HP:0003363 Previous pregnancy: TOP for heterotaxia and complex heart defect	VUS <i>DNAH11</i>	COP Live birth (41)	No	M24 No additional finding
Patient 74	No	30	Intrauterine growth retardation HP:0001511 Microcephaly HP:0000252 Short fetal femur length HP:0011428 Talipes equinovarus HP:0001762 Single umbilical artery HP:0001195	negative	COP Live birth (35)	Poor suck HP:0002033 Anemia HP:0001903 Alcohol, tobacco and opiates withdrawal	Clinical follow-up not requested by parents
Patient 78	Yes	27	Intrauterine growth retardation HP:0001511	negative	COP Live birth (38)	No	M24 Microcephaly HP:0000252 Strabismus HP:0000486 Neurodevelopmental delay HP:0012758
Patient 79	No	10	Fetal cystic hygroma HP:0010878 Hydrops fetalis HP:0001789	negative	COP Live birth (41)	No	M17 No additional finding
Patient 84	No	20	Intrauterine growth retardation HP:0001511 Unilateral renal agenesis HP:0000122	negative	COP Live birth (32)	No	M22 Esophageal atresia HP:0002032, Delayed speech

			Single umbilical artery HP:0001195				and language development HP:0000750 Anal atresia HP:0002023
Patient 85	No	20	Polyhydramnios HP:0001561 Abnormal corpus callosum morphology HP:0001273 Macrocephaly HP:0000256 Interhemispheric cyst HP:0032327	negative	COP Live birth (34)	Large for gestational age HP:0001520	Neonatal death Brain neoplasm HP:0030692
Patient 86	No	20	Intrauterine growth retardation HP:0001511 Polyhydramnios HP:0001561 Microcephaly HP:0000252 Abnormal facial shape HP:0001999	negative	COP Live birth (26)	No	M26 Strabismus HP:0000486 Hypermetropia HP:0000540 Asthma HP:0002099 Sucking-swallowing disorders Neurodevelopmental delay HP:0012758
Patient 87	No	23	Cerebellar vermis hypoplasia HP:0001320 Abnormal cortical gyration HP:0002536	VUS <i>DLL1</i>	COP Live birth (26)	No	M12 Motor delay HP:0001270
Patient 94	No	20	Agenesis of corpus callosum HP:0001274 Enlarged fetal cisterna magna HP:0011427 Bicuspid aortic valve HP:0001647 Lissencephaly HP:0001339	VUS <i>KMT2E</i>	TOP (30)	Macrocephaly HP:0000256 Hypertelorism HP:0000316 Microretrognathia HP:0000308 Low-set ears HP:0000369	NA
Patient 96	No	20	Large for gestational age HP:0001520 Pierre-Robin sequence HP:0000201 Abnormal thymus morphology HP:0000777	negative	COP Live birth (33)	Aplasia of the thumb HP:0009601	M22 Sucking-swallowing disorders
Patient 97	No	20	Hypoplasia of the corpus callosum HP:0002079	negative	COP Live birth (40)	Agenesis of corpus callosum HP:0001274 Iris coloboma HP:0000612 Aplasia of the ear HP:0008771	M20 Micrognathia HP:0000347 Hypoplasia of the semicircular canal HP:0011382

Patient 98	No	19	Bladder exstrophy HP:0002836 Anal atresia HP:0002023	negative	COP Live birth (39)	No	M24 No additional finding
Patient 100	No	20	Agenesis of corpus callosum HP:0001274 Short fetal femur length HP:0011428	negative	COP Live birth (34)	No	M29 Delayed ability to sit HP:0025336 Sucking-swallowing disorders
Patient 101	No	30	Agenesis of corpus callosum HP:0001274	negative	COP Live birth (39)	No	M24 No additional finding
Patient 103	No	21	Ureteral duplication HP:0000073 Talipes equinovarus HP:0001762 Single umbilical artery HP:0001195	negative	COP Live birth (38)	No	Clinical follow-up not requested by parents
Patient 109	No	10	Fetal cystic hygroma HP:0010878 ? Ventricular septal defect HP:0001629 Short fetal femur length HP:0011428	negative	COP Live birth (41)	No Ventricular septal defect HP:0001629	M24 No additional finding
Patient 112	No	20	Short long bone HP:0003026 1-2 toe syndactyly HP:0010711 Abnormal male external genitalia morphology HP:0000032	negative	COP Live birth (33)	Cryptorchidism HP:0000028 Hypospadias HP:0000047	Clinical follow-up not requested by parents
Patient 115	No	20	Tetralogy of Fallot HP:0001636 Fetal pyelectasis HP:0010945	negative	COP Live birth (38)	No	M15 No additional finding
Patient 117	No	20	Abnormal facial shape HP:0001999 Long philtrum HP:0000343 Ectopic kidney HP:0000086 Short fetal femur length HP:0011428	negative	COP Live birth (41)	No	Clinical follow-up not requested by parents
Patient 123	No	20	Tetralogy of Fallot HP:0001636 Abnormal male external genitalia morphology HP:0000032	VUS KAT7	TOP (26)	Micropenis HP:0000054 Low-set ears HP:0000369 Retrognathia HP:0000278 Short philtrum HP:0000322	NA

						Hypertelorism HP:0000316 Long eyelashes HP:0000527 Sloping forehead HP:0000340 Single umbilical artery HP:0001195 Microcornea HP:0000482	
Patient 122	No	30	Preauricular skin tag HP:0000384 Congenital portosystemic venous shunt HP:0012022 Fetal pyelectasis HP:0010945 Hypospadias HP:0000047	negative	COP Live birth (37)	Ureteral duplication HP:0000073 Ventricular septal defect HP:0001629 Sacral segmentation defect HP:0008490 Hearing impairment HP:0000365 Anorectal anomaly HP:0012732	M13 No additional finding
Patient 125	No	20	Intrauterine growth retardation HP:0001511 Atrial septal defect HP:0001631 Ventricular septal defect HP:0001629 Abnormal thymus morphology HP:0000777 Single umbilical artery HP:0001195	negative	COP Live birth (34)	Bicuspid aortic valve HP:0001647 Microcephaly HP:0000252 Asymmetry of the mouth HP:0009941 Intrauterine growth retardation HP:0001511	Clinical follow-up not requested by parents
Patient 126	No	20	Cleft lip HP:0410030 Occipital meningocele HP:0002436	negative	COP Live birth (35)	No	M12 Rhombencephalosynapsis HP:0031913 Subcortical heterotopia HP:0032391
Patient 130	No	20	Congenital diaphragmatic hernia HP:0000776 Dextrocardia HP:0001651 Ectrodactyly HP:0100257	negative	COP Live birth (38)	Syndactyly HP:0001159	M12 Pulmonary arterial hypertension HP:0002092 Hypertonia HP:0001276
Patient 131	No	21	Oligohydramnios HP:0001562 Ventricular septal defect HP:0001629 Renal agenesis HP:0000104	negative	IUFD (29)	Median cleft palate HP:0009099 Short neck HP:0000470	NA

			Talipes equinovarus HP:0001762			Abnormal facial shape HP:0001999 Hypoplasia of the thumb HP:0009601 Congenital diaphragmatic hernia HP:0000776 Abnormal mesentery morphology HP:0100016 Single umbilical artery HP:0001195 Cerebellar vermis hypoplasia HP:0001320 Hemivertebrae HP:0002937 Polysplenia HP:0001748 /Hypoplasia of the scapulae HP:0006713 Missing ribs HP:0000921 Nodular neuronal heterotopia ORPHA:2149 Abnormal uterus morphology HP:0031105	
Patient 132	No	20	Short corpus callosum HP:0200012 Median cleft lip and palate HP:0008501	negative	COP Live birth (38)	No	NA
Patient 134	No	20	Intrauterine growth retardation HP:0001511 Ambiguous genitalia, male HP:0000033	negative	COP Live birth (30)	No	M21 No additional finding
Patient 136	No	20	Dolichocephaly HP:0000268 Broad neck HP:0000475 Polycystic kidney dysplasia HP:0000113 Echogenic fetal bowel HP:0010943	negative	COP Live birth (39)	No	M13 No additional finding
Patient 139	No	20	Hemivertebrae HP:0002937 Talipes equinovarus HP:0001762	dupXq28	TOP (36)	No (postmortem examination not accepted)	NA

			Lateral ventricular asymmetry HP:0100960 Dysostosis multiplex HP:0000943				
Patient 141	No	20	Agenesis of corpus callosum HP:0001274 ? Interhemispheric cyst HP:0032327	negative	TOP (31)	No (postmortem examination not accepted)	NA
Patient 142	No	20	Polyhydramnios HP:0001561 Abnormal heart morphology HP:0001627 Single umbilical artery HP:0001195	negative	COP Live birth (38)	Mesocardia HP:0011599, Atrial situs ambiguous HP:0011539	Neonatal death
Patient 145	No	20	Agenesis of corpus callosum HP:0001274	VUS <i>BRD2</i>	TOP (33)	No (postmortem examination not accepted)	NA
Patient 147	No	20	Agenesis of corpus callosum HP:0001274	negative	COP Live birth (37)	No	M17 No additional finding

COP: Continuation of pregnancy, TOP: termination of pregnancy