Case number	**Gestational age at** MR **(weeks+days)**	**Nature of GE abnormality (B = bilateral, U =unilateral, C= cavitation, E = enlargement)**	**FOD centile for GA (mean +/- n SD)**	**Pregnancy outcome and results of genetic testing (TOP = termination of pregnancy)**	**Clinical outcome of liveborn subjects (n/a=not applicable)**
1	22+5	UE	+5 SD for overgrown hemisphere. +1 SD for contralateral hemisphere	TOP. No genetic testing. Presumptive diagnosis of TSC - related hemispheric overgrowth syndrome in view of cardiac rhabdomyoma on prenatal US.	n/a
2	27+6	UE	+6 SD	TOP. c. 2176G>A pathogenic variant in *PIK3CA* gene	n/a

3	22+6	BE	+7 SD	TOP. *MTOR* c.6644C>Tp. (Ser2215Phe) mosaic pathogenic variant	n/a
4	25+1	UE. Haemorrhage within enlarged GE extending into hemisphere.	+3 SD	No aneuploidy. Genomic testing not performed.	Hemimegalencephaly. Intractable seizures. Functional hemispherectomy at 8/12. No genomic testing on excised tissue. Developmental delay.
5	25 + 1	BE	+6 SD	TOP. *De novo* pathogenic variant in the *MTOR* gene c.5930_5931del A>G; p.Thr1977Lys. This variant is known to be associated with focal cortical dysplasia, hemimegalencephaly and polymicrogyria and a diagnosis	n/a

				of Smith-Kingsmore syndrome (OMIM # 616638).	
6	31+4	BE	+4 SD	No genomic testing in view of clinical and imaging phenotype suggesting mTOR / *PIK3CA* overgrowth syndrome	Overlapping features of megalencephaly capillary malformation (MCAP) and megalencephaly polydactyly polymicrogyria hydrocephalus (MPPH). Postaxial polydactyly in both feet and facial capillary malformation. Seizures began at 7 /12 postnatal with epileptic discharges left occipitotemporal region. Behavioural and learning problems.
7	30+3	UE	+1 SD	*TSC2* pathogenic copy number variant diagnosed on prenatal microarray (details of the variant	MR appearances diagnostic for tuberous sclerosis. Epilepsy,

				not available). Father known to have tuberous sclerosis.	developmental delay, renal angiomyolipoma
8	25 +2	UE	+2 SD	Not performed. Father has a clinical diagnosis of tuberous sclerosis.	MR appearances diagnostic for tuberous sclerosis. Epilepsy and developmental delay
9	23+2	BC	-3 SD	TOP. Trio WES demonstrated homozygous variants of unknown significance in *VARS*, *VARS2 and CUL7* genes. The parents are consanguineous and are carriers of these variants. Unclear if one or more of these variants contributed to the phenotype in the affected fetuses in this family.	Prenatal MR findings typical for Fetal Akinesia Dyskinesia Sequence (FADS).

10	23+3	BC	-2SD	Trio WES identified a *de novo* pathogenic *PDHA1* variant in child: NM_000284.3, c.904>T, p.Arg302Cys).	Severe cerebral palsy. Currently 6 years of follow up; multiple admissions for aspiration pneumonia. Severe epilepsy from 12/12 of age.
11	22+2	BE	-2.5 SD	Heterozygous de novo pathogenic variant *TUBA1A* gene (c1265G>A).	Seizures, severe developmental delay. 1 year and 10 months of follow up; multiple admissions for aspiration pneumonia.
12	24+1	BE, BC	-2 SD	Neuromuscular disease gene panel performed*. Multiple sequence variants detected but none considered clinically relevant.	Walker Warburg clinical and imaging phenotype with cobblestone lissencephaly. Deceased at 63 days of age. Seizures, respiratory failure and poor feeding; poor tone.
13	22+6	BE	-3 SD	TOP. Heterozygous *de novo* pathogenic variant identified in	n/a

				TUBA1A gene on trio exome. Variant details unavailable.	
14	23+0	BE	-2 SD	Heterozygous *de novo* pathogenic variant c.74G>T (p.Cys25Phe) in exon 2 of *TUBA1A* gene.	n/a
15	22+4	BE, BC	-3 SD	n/a	Cobblestone lissencephaly, Walker Warburg phenotype on genetics review and imaging. Death at 4 months of age due to poor tone / swallowing / respiratory failure.
16	20+3	BE	-3 SD	TOP. Heterozygous *de novo* pathogenic variant *TUBA1A* c.719C>G (p.Ala240Gly) on WES. Initial gene panel testing in 2014 for 5 genes known to	Cobblestone lissencephaly at post-mortem. Elder sibling of Subject 17.

				cause Walker - Warburg syndrome was negative.	
17	21+1	BE	-2 SD	Heterozygous *de novo* pathogenic variant in the *TUBA1A* gene c.719C>G (p.Ala240Gly). Germline mosaicism in parent suspected but unconfirmed due to recurrence of variant in two siblings.	Neonatal death (day1). Younger sibling of Subject 16.
18	24+4	BE	-2.5 SD	TOP. Heterozygous *de novo* pathogenic missense variant *TUBAIA*. OMIM # Lissencephaly 3.611603 AD.	n/a

19	33 + 0	BC	- 2 SD	Heterozygous *de novo* pathogenic *TUBA1A* c.887T>G (p.Phe296Cys)	Developmental delay, visual impairment, hypoacusis, microcephaly, epilepsy.
20	26 + 5	BE	-2SD	TOP. Heterozygous *de novo* pathogenic variant *TUBB3* c.1138C>T;p.Arg380Cys	n/a
21	31 +0	BE	0 SD (at the mean)	*Heterozygous x – linked pathogenic variant OPHN1* pathogenic mutation. deletion 3020 bp: NG_008960.1 g.242351_245371. Deletion of exon 13 e 14 RNA mess (NM_002547.2 c.1105_1201del) frameshift mutation and premature stop codon (NP_002538.1 p.IIe369 fs*21).	Febrile seizures. Mild neurodevelopmental delay.

				Same deletion found in one X chromosome of the mother.	
22	22 +3	BG, BC	-0.5SD	Blood and skin microarray - normal. DNA extracted from biopsy of Blashkoid depigmented skin lesion (dark-skinned infant) failed to demonstrate explanatory mutation on WES.	Seizures including infantile spasms, visual impairment, developmental delay, cerebral palsy.

Table 1. Study subjects: head size and clinical, genetic and pathologic diagnoses

Legend:

AD = autosomal dominant

a/a = not applicable

SD = standard deviations

US = ultrasound

WES = whole exome sequencing

SD = standard deviations

TOP = termination of pregnancy

*This included a neuromuscular subexomic supercapture of over 400 genes known to be associated with ataxia, congenital muscular dystrophy, mitochondrial disease, spinal muscular atrophy, myopathies, limb girdle muscular dystrophies, lissencephalies (including TUBA1A and TUBB3 but no other tubulin genes) and distal arthrogryposis.

	Case number	GA at US preceding first MR (weeks + days)	Pre MRI US Findings	GA at first MR (weeks+days)	FOD centile for GA	Nature of GE abnormality (B = bilateral, U = unilateral, C= cavitation, E = enlargement)	Hemispheres	Cerebellum	Brainstem	Corpus callosum	GA at second MR	Second MR findings	GA at third MR	Third MR findings	Amniocentesis (1= Yes; 0 = No)	Amniocentesis result	Pregnancy outcome	GA at delivery (weeks and days)	Postnatal MR (Y/N)	Age at postnatal MR (days).	Postnatal MR findings	Postmortem findings (n/a = not applicable)	Genetic testing 1=performed, 0=not performed	Results of genetic testing	Clinical outcome of liveborn (n/a=not applicable)
	1	12+4	DCDA twins. One twin has cardiac left ventricular mass, presumed rhabdoymoma and mass lesion in one cerebral hemipshere - possible haemorrhage or tumor? Ipsilateral VM 12mm.	22+5	5 SD above the mean for overgrown hemisphere. 1 SD > mean for contralateral hemipshere	UE	Overgrowth of hemisphere ipsilateral to enlarged GE. AbN opercularisation.	N	N	dysgenetic - abnormal shape	24+5	Twin 1- extensive malformation of cortical development, cardiac mass, most likely tuberous sclerosis complex, mass like component probable hamartoma Twin 2 - N	n/a	n/a	0	n/a	Selective TOP Twin 1 Liveborn N Twin 2	34+2	no	np	n/a	np	0	No genetic testing. Presumptive diagnosis of TSC - related hemispheric overgrowth syndrome in view of prenatal MR and US findings.	n/a
	2	23+6	Large for dates fetus, bilat VM right ventricle 14.1mm left ventricle 11.9mm	27+6	6 SD > mean	UE	Bilateral abN opercularization with perisylvian polymicrogyria. Bilateral mild lateral ventriculomegaly. Asymmetric enlargement of left hemisphere with posterior quadrant predominance and herniation of occipital lobe across midline.	N	N	N	n/a	n/a	n/a	n/a	1	N	TOP	30+2	no	np	n/a	Megalencephaly, polymicrogyria, polydactyly both hands and left foot	1	c. 2176G>A pathogenic variant in PIK3CA gene	n/a
	3	22+6	Macrocephaly with ventriculomegaly of 12mm, absent cavum septum pellucidum, likely absent corpus callosum	22+6	7 SD > mean	BE	Mild bilateral ventriculomegaly. Intrav	N appearance but transverse cerebellar diameter and vermis height 2SD above mean for GA	N appearance but AP diameter pons 2 SD above mean for GA	N	26+6	Bilateral perisylvian polymicrogyria, abnormally enlarged bilateral ganglionic eminences, cavitation no longer evident, and disorganised appearance to periventricular crossroads. Persistent megalencephaly.	n/a	n/a	0	n/a	TOP	37	n/a	n/a		Macrocephaly, bilateral hemispheric polymicrogyria, dysplasia of the thalami, fragmentation of hindbrain due to postmortem change.	1	MTOR c.6644C>Tp. (Ser2215Phe) pathogenic mosaic variant	n/a
	4	20+2	Hemimegalencephaly	25+1	3 SD > mean	UE. Small focus of haemorrhage centrally within enlarged GE extending into hemisphere.	Asymmetric overgrowth of one hemisphere.	N	N	N	30+1	Right hemimegalencephaly. Resolution of right parenchymal haemorrhage with porencephalic cyst formation.	n/a	n/a	0	n/a	liveborn	39+3	yes	4	Right hemimegalencephaly. Bilateral germinal matrix haemorrhage. No malformation of the left cerebral hemisphere	n/a	1	No aneuploidy. Whole exome sequencing not performed.	Intractable seizures. Functional hemispherectomy at 8/12. Developmental delay
	5	22+1	Cranial biometry >95th percentile. Agenesis of corpus callosum.	25 + 1	6 SD > mean	BE	BE left greater than right	N	N	Length 2 SD < mean for GA	n/a	n/a	n/a	n/a	1	N microarray	TOP	27+6	N	np	n/a	np	1	Pathogenic variant in the MTOR gene c.5930_5931del A>G; p.Thr1977Lys. This change is known to be associated with focal cortical dysplasia, hemimegalencephaly and polymicrogyria and a diagnosis of Smith-Kingsmore syndrome (OMIM # 616638).	n/a
	6	28+2	Hydrocephalus, echogenic bowel, polyhydramnios	31+4	4 SD > mean	BE	Asymmetric overgrowth of one hemisphere. Bialteral abnormal opercularisation and perisylvian polymicrogyria	Morphologically N. Vermis height 6 SD and transverse cerebellar diameter 3 SD above mean for GA	N. AP pontine diameter at the mean for GA	N	n/a	n/a	n/a	n/a	1	N	liveborn	39+3	yes	6	Left hemispheric hemimegalencephaly with posterior quadrant predominance, extensive left cerebral hemisphere polymicrogyria, , mild Bilateral ventricular dilatation, arachnoid cyst left anterior temporal pole. Lipoma superior to right cerebellar hemisphere, overgrowth of fat left greater than right both premaxillary areas Ventricular shunt placement at 12/12 for obstructive hydrocephalus.	n/a	1	No genetic diagnosis testing in view of phenotype suggesting mTOR / PI3KCA overgrowth syndrome	Seizures documented at 7 /12 postnatal with epileptic discharges left occipitotemporal region. Behavioural, learning problems. Overlapping features of megalencephaly capillary malformation (MCAP) and megalencephaly polydactyly polymicrogyria hydrocephalus (MPPH). Postaxial polydactyly in both feet and facial capillary malformation diagnosed postnatally.
	7	25+3	Left ventricular cardiac rhabdomyoma. N intracranial exam.	30+3	1 SD > mean	UE	Multiple small hemispheric mass lesions and subependymal nodules consistent with hamartomas and tubers.	N	N	N	n/a	n/a	n/a	n/a	1	Single copy of the c.4375C>T mutation in the TSC2 gene	liveborn male	42+0	yes	177	MRI at 6/12 shows typical features of TSC.	n/a	1	TSC2 mutation diagnosed antenatally, father also known to have tuberous sclerosis	Epilepsy, developmental delay, renal angiomyolipoma
	8	21	2 cardiac left ventricular rhabdomyomas	25 +2	2 SD > mean	UE	N apart from bilateral mild (11mm) ventriculomegaly	N	N	N	n/a	n/a	n/a	n/a	0	NA	liveborn male	41+0	yes	5	Head MRI day 5 of life shows typical TSC.	n/a	0	Not performed. Father has tuberous sclerosis also.	Epilepsy and developmental delay
	9	20	Severe VM bilaterally. Small cerebellum. Prior TOP for fetal brain abnormalities, consanguinity.	23+2	3 SD < mean	BC	Thin parenchyma, underopercularization, severe VM.	3SD below mean for transverse diameter and vermis biometry	Thin	Abnormally thin - hypogenetic	n/a	n/a	n/a	n/a	1	N	TOP	24+0	n/a	n/a	n/a	n/a	1	Trio WES demonstrated 3 variants of unknown significance involving genes on chromosome 6; the fetus was biallelic for the variants and each parent was a heterozygous carrier of the variant. These were: ***VARS*** chr6(GRCh38): g.31784478G> A NM_006295.2: c.1492C>T p.(Arg498Cys) ***VARS2*** chr6(GRCh38): g. 30918868C>T NM_001167734.1 c.1027C>T ***CUL7*** chr6(GRCh38): g.43051758G>A NM_001168370.1 c.586C>T p.{Arg196Trp) p.(Pro343Ser)	MR findings typical of Fetal Akinesia Dyskinesia Sequence (FADS). Parents are consanguinous.
	10	21+3	Dysgenesis CC, hypertelorism, colpocephaly	23+3	2SD < mean	BC	Bilateral mild (11mm) ventriculomegaly. Bilateral anterior temporal pole cysts.	Transverse cerebellar diameter 3 SD < mean. N vermis	N	ACC. Anterior commissure present	n/a	n/a	n/a	n/a	1	N microarray	liveborn	41+6	yes	31	ACC, with severe loss of periventricular white matter mimicking prematurity - related periventricular white matter injury.	n/a	1		Severe cerebral palsy.Currently 6 years of follow up with multiple admissions for aspriation pneumonia. Severe epilepsy from 12/12 of age.
	11	21+2	Abnormal appearance of posterior fossa, cystic structure - arachnoid cyst or Blake's pouch? Probable ACC.	22+2	Between 2 and 3 SD below mean	BE	N	Persistent Blake's pouch cyst with rotated vermis. AP diameter of vermis 2SD below mean. Transverse cerebellar diameter on 50th centile but artificially increased by dilated 4th ventricle displacing the cerebellar hemispheres laterally.	N	ACC with interhemispheric cyst. Bilateral mild copocephaly 10mm and 11mm atrial diameter.	28+0	ACC, persistent Blake's pouch, vermis and pontine biometry below 3rd percentile, sulcation moderately delayed for GA	36+0	Persistent FOD <3rd centile, ACC, abnormal enlargement of the ganglionic eminences. Bilateral abormality of opercularization, hemispheric dysgyria. Pontine AP diameter 2SD below mean for GA and asymmetric mid pons on axial images. Persistent dilated Blake's pouch with vermis rotation and persistent reduced AP vermis diameter. Constellation of findings highly suggestive of tubulinopathy.	1	N microarray	liveborn	41+2	yes	1	ACC. Enlarged basal ganglia with absent anterior limb internal capsule. Bilateral abN opercularisation and extensive dysgyria. Brainstem dysplasia with small pons and asymmetry of ventral pons on axial images.Vermis upwardly rotated by persistent Blake's pouch. Hypogplasia of vermis and cerebellar hemispheres. Findings typical for tubulinopathy.	n/a	1	Heterozygous for c1265G>A pathogenic variant in TUBA1A gene	Seizures, severe developmental delay. 1 year and 10 months of follow up with multiple hospital admissions for aspiration pneumonia.
	12	20/40	ACC. Persistent Blake's pouch.	24+1	2 SD below mean	BE, BC	Marked underopercularization. Parietooccipital sulcus and cinglulate sulcus not seen. Severe lateral ventriculomegaly.	Transverse cerebellar diameter 2 SD below mean for GA	Thin and kinked. Deep ventral pontine cleft on axial images. AP pontine diameter 3SD below mean for GA.	ACC	32+2	Walker Warburg phenotype, cerebellar hypoplasia affecting vermis and hemispheres, z-shaped kinking of thin brainstem ,thin hemipsheric parenchyma with absent sulcation, ACC, ventral pontine cleft, bilateral severe VM	n/a	n/a	1	N microarray	liveborn	40+1	yes	7	Cobblestone lissencephaly, ACC, kinked thin brainstem, cerebellar hypoplasia, enlarged dysmorphic basal ganglia. Consistent with Walker Warburg phenotype.	no PM	1	Neuromuscular disease gene panel analysis-multiple sequence variants detected but none considered clinically relevant. Tubulin genes not included in tested panel	Deceased at 63 days of age. Principal diagnoses-lissencephaly, seizures, respiratory failure related to poor tone, neurological feeding problems, diabetes insipidus, hypernatraemia, sepsis
	13	22+1	Microcephaly, bilateral ventriculomegaly left -11mm, right -14mm), CSP and CC not identified, cerebellum small with no obvious vermis, brainstem small with flattening of pons	22+6	3 SD below mean	BE	Thin parenchyma, underopercularization, moderately severe lateral ventriculomegaly (13mm) . Subependymal nodules lining one frontal horn.	3 SD below mean for GA (hemispheres and vermis)	Thin. AP diameter of pons 3 SD below mean for GA.	ACC	n/a	n/a	n/a	n/a	1	N microarray	TOP	23+3	no	np	n/a	Facial dysmorphism, microcephaly, underdeveloped sulci, ventriculomegaly, ACC, hypoplastic cerebellum	1	De novo pathogenic variant TUBA1A gene. Both parents are negative for the mutation, therefore variant reclassified as pathogenic	n/a
	14	22+2	Severe VM, 16mm atrial diameter bilaterally, echogenic material, consistent with clot within ventricles, small cerebellum, CSP present but thin CC noted	23+0	2 SD below mean	BE	Underopercularization bilaterally. Bilateral VM (16mm and 13mm). Thin hemispheric parenchyma.	Vermis height and transverse cerebellar diameter 2 SD below the mean	N	Present and complete but abormally thin	25+0	Increased lateral VM now 15-16mm, hypogenic CC, failure of progression of primary sulcation since previous MR, continued cerebellar hypoplasia	n/a	n/a	0	n/a	TOP	25+2	no	np	n/a	severe brain maceration, absent CC, dilated ventricles	1	heterozygous variant c.74G>T (p.Cys25Phe) in exon 2 of TUBA1A gene	n/a
	15	21+3	Bilateral cerebral ventriculomegaly 13 mm. Suspected haemorrhage into the choroid plexus bilaterally	22+4	3 SD below mean	BE, BC	Severe ventriculomegaly and underopercularization.	Vermis and transverese cerebellar diameter 2SD below mean	Not kinked. Pons AP diameter 1SD below mean for GA	ACC	n/a	n/a	n/a	n/a	1	N	Liveborn female	39+2	Y	63	Severe ventriculomegaly. Cobblestone lissencephaly	n/a	0	n/a	Death at 4 months of age due to poor tone / respiratory failure / poor swallowing
	16	19+3	Small cerebellum	20+3	3 SD below mean	BE	Bilateral moderately severe VM. Thin cerebral parenchyma.	Reduced cerebellar and vermis diameters 3 SD below mean	Mild pontine kink. Pons AP diameter 2 SD below mean	ACC	n/a	n/a	n/a	n/a	1	n/a	TOP	23+5	N	n/a	n/a	cobblestone lissencephaly	1	heterozygous variant in the TUBA1A gene c.719C>G (p.Ala240Gly). However gene panel testing in 2014 for 5 genes known to cause Walker - Warburg syndrome was negative.	Elder sibling of 2.3
	17	19+0	Moderate ventriculomegaly, hypoplastic cerebellum, absent CSP, micrognathia	21+1	2 SD below mean	BE	No significant opercularization. Thin hemipsheric parenchyma. Severe bilateral lateral VM	Reduced cerebellar and vermis diameters 3 SD below mean	Mild pontine kink. Pontine AP diameter 2 SD below mean for GA.	ACC	34+0	n/a	n/a	n/a	0	n/a	Liveborn	35+3	N	n/a	n/a	n/a	1	heterozygous variant in the TUBA1A gene c.719C>G (p.Ala240Gly) as for sibling. Germline mosaicism in parent suspected due to recurrence of AD somatic pathogenic mutation in two siblings.	Neonatal death (day 1). Younger sibling of 2.2.
	18	21+4	Posterior fossa abnormality. Dandy Walker malformation?	24+4	2.5 SD below mean for GA	BE	No significant opercularization. Severe bilateral VM.	Upward rotation of cerebellum consistent with persistent Blake's pouch. Vermis too small to be measured accurately. Transverse cerebellar diameter at 10th centile.	Thin, kinked. AP diameter of pons on 10th centile. Ventral pontine cleft.	ACC	n/a	n/a	n/a	n/a	n/a	n/a	TOP	n/a	N	n/a	n/a	n/a	1	Heterozygous missense mutation TUBAIA. OMIM # Lissencephaly 3.611603 AD.	n/a
	19	n/a	n/a	33 + 0	More than 2 SD below the mean for GA	BC	Underopercularization. Underdeveloped primary and secondary sulcation. Abornal persistence of laminated appearance. Bilateral VM > 12mm.	TCD and vermis height more than 2SD < mean. Severe global cerebellar hypoplasia	N	N	n/a	n/a	n/a	n/a	0	n/a	lLiveborn female	40	Y	n/a	Typical features of tubulinopathy - lissencephaly enlarged basal ganglia, absent anterior limb of internal capsule, cerebellar hypoplasia	n/a	1	Heterozygous TUBA1A c.887T>G (p.Phe296Cys)	Epilepsy
	20	26+5	Bilateral ventriculomegaly moderate right, severe left, hypoplastic pons, abnormal brain gyration and hypotelorism.	26 + 5	2SD <mean	BE	Abnormal opercularization, dysgyria. Moderate right and severe left ventriculomegaly.	Morphologically normal. Transverse diameter 13th centile for GA.	AP diameter of pons 10th centile for GA. No brainstem kink.	Present	n/a	n/a	n/a	n/a	n/a	n/a	TOP		N	n/a	n/a	n/a	1	Class 5 (pathogeic) TUBB3 variant c.1138C>T;p.Arg380Cys	n/a
	21	n/a	n/a	29 + 0	50th centile	BE	N apart from mild bilateral VM	N	N	N	n/a	n/a	n/a	n/a	0	n/a	liveborn male		Y	7 years	Periventricular nodular heterotopia. Persistent mega cisterna magna. Mesial temporal sclerosis.	n/a	1	Oligophrenin 1 mutation. deletion 3020 bp: NG_008960.1 g.242351_245371. Deletion of exon 13 e 14 RNA mess (NM_002547.2 c.1105_1201del) frameshift mutation and premature stop codon (NP_002538.1 p.Ile369 fs*21) . Same deletion found in one X chromosome of the mother.	Febrile seizures. Mild neurodevelopmental delay.
	22	22+3	Unilateral VM 12mm, difficulty visualising CSP.	25+1	30th centile	BG, BC	Bilateral abnormal opercularization and perisylvian polymicrogyria.	N	N	N	32+1	Subbstantial reduction in size of GEs compared with prior examination but still abormally large on the right, bilateral frontal subependymal pseudocysts, CC present but thinned. Abnormal opercularization persists. Sulcation has progressed appropriate for gestation.	n/a	n/a	0	n/a	liveborn	38+6	yes	58	Right frontal polymicrogyria, abormal orientation of Sylvian fissures, bilateral subependymal pseudocysts.	n/a	1	Blood and skin microarray - normal. Postnatal biopsy of Blashkoid depigmented skin lesion (dark - skinned infant) failed to demonstrate explanatory mutation on WES.	Seizures including infantile spasms, visual impairment, developmental delay cerebral palsy

LEGEND

ACC = agenesis of the corpus callosum
AP = anteroposterior
CSP = cavum septum pellucidum
CC = corpus callosum
GA = gestational age
N = Normal
n/a = not available or not applicable
np = not performed
TSC = tuberous sclerosis complex
VM = ventriculomegaly
WES = whole exome sequencing