

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

3	22+6	BE	+7 SD	TOP. <i>MTOR</i> 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. <i>De novo</i> pathogenic variant in the <i>MTOR</i> 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	developmental delay, renal
				have tuberous sclerosis.	angiomyolipoma
8	25 +2	UE	+2 SD	Not performed. Father has a	MR appearances diagnostic for
				clinical diagnosis of tuberous	tuberous sclerosis. Epilepsy and
				sclerosis.	developmental delay
9	23+2	BC	-3 SD	TOP. Trio WES demonstrated	Prenatal MR findings typical for Fetal
				homozygous variants of	Akinesia Dyskinesia Sequence
				unknown significance in VARS,	(FADS).
				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.	

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

				TUBA1A gene on trio exome. Variant details unavailable.	
14	23+0	BE	-2 SD	Heterozygous <i>de novo</i> pathogenic variant c.74G>T (p.Cys25Phe) in exon 2 of <i>TUBA1A</i> 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 <i>de novo</i> pathogenic variant <i>TUBA1A</i> c.719C>G (p.Ala240Gly) on WES. Initial gene panel testing in 2014 for 5 genes known to	Cobblestone lissencephaly at postmortem. Elder sibling of Subject 17.

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

	Developmental delay, visual
pathogenic TUBA1A c.887T>G	impairment, hypoacusis,
(p.Phe296Cys)	microcephaly, epilepsy.
TOP. Heterozygous de novo	n/a
pathogenic variant TUBB3	
c.1138C>T;p.Arg380Cys	
Heterozygous x – linked	Febrile seizures. Mild
pathogenic variant OPHN1	neurodevelopmental delay.
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).	
	(p.Phe296Cys) TOP. Heterozygous <i>de novo</i> pathogenic variant <i>TUBB3</i> c.1138C>T;p.Arg380Cys <i>Heterozygous x – linked</i> pathogenic variant <i>OPHN1</i> 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

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

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.

Cavanabo	ga.	A at US proceeding first MR (works + days) Pro MRI US Findings	GA at foot h	MR N) FOD consile for GA	Nature of GE abnormality (R = bilinard, U stellature), C = contation, E = solvegement)	:- Hemispheres	Controllars Brainstan Corres o	GA at second tallocate MR	Guond MR Godines	arthind MR Third MR fedines	Aminomenia (1-Yes; 0	Promance outcome	GA at Salivary Weeks and Sans) Postmatal MR (Age at postnatal MR	Percusal MR findees	Guaric tenting 1 "performed, 0"-net performed to the continual of the cont	Results of emotic tertifier	Clinical outcome of Ecohom — to invast assistable)
		DCDA twiss. One twin has recorded shabdowness and	medio left vonticular mass, mass lecion in one-carabtal obage or tumor? Spelatonal 22s.5						Twin 1-extensive malformation of cortical development, cardiac								No sensis torine, Procurerive damonic of TSC - related	
		hamipshara - poscible hauso 12=4 VM 12mm	nhage or tumor? Spolatonii 22+5	55D above the mean for overgrown hamisp 15D > mean for contributed hemipshare	Phone. UE	Overgrowth of homisphere ipolatical to enlarged GE. AbN openiclarication.	N N Apageneti	tic - absormal shape 24-5	Twin 1- occursive multi-transition of cortical development, cardiac mass, most likely subsects submosic complex, mass like component probable humatoms. Twin 2 - N	6/4		Saluctive TOP Twin 1 Liveborn N Twin 2	14-2 00	*	ab.		No genetic testing. Procumptive diagnosis of TSC - related beningbasic overgrowth syndrome in view of general MSR and US findings.	eh.
						Bilanci abN operatazization with paricylvian polymicrogyvia. Bilancial												
		Large for dates fatus, bila V	M right vontricle 14 Imm left			Ellistaral abN opensularization with parity/vian polyminosgyvia. Ellistaral stild latural visiticalismoguly. Asymmetric unlargenance of left lasticiphers with poeterier quadrant professioness and homistion of control laboratories reference.										Magakeusphaly, polymicogysia, polydanyly both hands and left	A NAME A AND ADDRESS OF THE PARTY OF	
		279	2,79			coupies and across means.			**							1	a. 2176G>A pathogosic variant in PIKICA gene	
									Bilatural paricylvian polymicrogyvia, abnormally enlarged									
,		Macrocophily with vanticul arous coptum policidus, li	omogaly of 12mm, about toly about corpus collocum 22+6	75D>mm	GE.	Mild bilased conticulonogaly: Intrav	N appearance but transverse corrboller diameter and version leight 25D above mean for GA. above mean for GA.	26+6	Bilatural paricylvian polymicrogyvia, abasemally eslanged bilatural gauginoise essionesse, curitation no benger evident, and divergament appearance to polymontolicular concernade. Persistent magaine-upholy.	nia nia	9 49	TOP	37 69	**		Macrocophily, bilateral hemispheric polymicrogyris, dysplania of the thelani, fragmentation of hindbrain due to postmertum change.	MTOR c.6644C>Tp. (Sur22159bs) pathogonic monsic variant	aia.
		20-2 Hominogalmosphaly	25+1	35D > man	UE. Small focus of harmonthage centrally within enlarged GE extending into homical	ocymmetric overgrowth of one hore. bossisphere.	N S	30-1	Right homimogalincophaly. Resolution of sight parenchymal humanithage with permosphalic cycl formation.	n/a	***	Sirahora	18+3 yes		Eight huminogskeouphaly. Ethanol garminal numic hasmorthage. No malformation of the left carebral humiophere	1	No anosphisty. Whole stress sequencing not performed. Pathogonic variant in the MTOR game a \$100_5931 dal A=G _i p.The 1971.[pc. This change in known to be accordant with food cortical depethods, homitogathoroghy and polymeropin and a diagnosis of Smith-Eingenmon eyndrome (OMIM # 616630).	Intractable solution. Functional hamisphenictomy at \$12. Developmental dulay
-		Consid biometry > 16th perce 22+1	nalle. Agenosis of corpus	65D > man	as.	BE left greater than right	N N Emph 2	SD < mon for GA als	aŭ.	nh i	g 1 N microscopy	TOP	17+6 N		ala.		cortical dysplania, huminogaloscophaly and polymicrogystis and a diagnosis of Smith-Kingermon syndrome (OMIM # 616678).	ata Solomos documented at 7/12 postnatal with solomic
						Acommunic response that co-	Marehologically N. Vernis								Left homispharie hominogaluncophaly with porturior quadrant prodominance, octomeries left controlal homisphare polymicogytia, sadd Bilancal ventricular dilutation, anotherial cycle fit annexis returned pole. Exposus experies or apide condular homisphere, severgeous de of fat left gentre than right both pomus illusy areas.			Subtress documented at 7/12 postnatal with updaptic discharges left ecciphotamperal negion. Bulturinaral, lauring problems. Overlapping finances of sugglinoupdaly apillary andiformation (MCAP) and sugglinoupdaly polyslaurity polysiarraypria. In photocapitals on LMFHs. Personal popularapy in host of photocapitals on LMFHs. Personal popularapy in host of test and facile supillary multisensation diagnosal
		28-2 Hydrocephalas, adogenic be	real, polyhydramios 21+4	45D > mass	as.	Asymmetric overgrowth of one busisphere, Stalment absorbed opercularization and paritylvian polyminografia Multiple small homospheric mass. Indices and subspendyman loodings annionment with homospheric mass.	beight 6 SD and transverse St. AP positive carefully SD above diseases at the mean fee GA.	ah.	aù.	**	a IN	litabana 2	19+3 yes		temporal pole. Espona superior to right conduction humaphous, averageous to of fat left greater than right both personalitary areas. Vanticular shout placement at 1312 for obstruction hydrocophalus.		No genetic diagnosis testing in view of phenotype suggesting n/TOR PERCA overgreeth syndrome	by-decophalis (MPFH). Pertantal polydactyly in both fact and facial capillary multicreation diagnosed portnatally.
		Left vermicular cardiac shabd 25=3 scare.	benyoma N intramaial	15D > man	CE	Multiple small homispheric mass lacious and subspendymal nodulos consistent with homestones and tabars.					Single copy of the s4079C>T munifon in the TSC2 game	Einsborn male	Q-0 ma	177	MRI at 612 shows torical features of TSC.		TSC2 matrice diagnosal astrustelly, father also known to have talescent editories	Enilares, developmental delay, ranal ancions video man
		21 2 cardiac left ventricular shab	disayunus 25+2	25D>man	CE.	N apart from bilanceal mild (1 lmm) contriculomogally	N N	ab .	ah.	**	4 NA	lirabora mala	11+0 yes		Hand MRI day 5 of life shows typical TSC.	6	Not performed. Either has believe a classic also	Epilopsy and developmental dulay
																	This WES demonstrated 3 variants of unknown significance involving game on demonstrate, the finite was helded for the variant and subjection was heavy process solid of the variant and subjection was heavy process solid of the variant and process solid process of the process of the process of \$4.0000 (\$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.0000 \$1.	1
																	p(App89Cys) FARE2 date(GRCh89); g-3893868C>TNM_001167734.1 c.1023C>T CULT date(GRCh89); e-48651286C>A NM_00116978.1	
																	LS86>T ρ(Ag196Tsp) ρ(Pm3-GSar)	
		Sover VM bilatanily. Small 20 Small brain also emailities, con	conhellers. Prior TOP for congressly. 23-2	15D < man	ac	This parenchyme, anderopercularization, severe VM.	25D below mean for transverse classifier and varies biometry. This Absorber	ally this - hypogenetic as le	eis .	na na	18	toe	04+0 wa	**	ab.	ab I		MR findings typical of Fetal Akinosia Dyskinosia Suquence (FADS). Pursum are consuguinous.
10		21+3 Dyugmenis CC, hyperuloris	m, colpocaphaly 23=3	26D < mon	ac	Stituted mild (11 mm) contriculomogaly. Stituted america tamporal pole cysts.	Transvarse combellar Gammar 3 SD < mean. N vermis N ACC. As	nhalor comminum prosunt sils	ah.	ab.	h I N microstroy	linabora.	83+6 yes	31	ACC, with severe less of pericenticular white matter minicking postutority - related pericenticular white nature injury.	ab I		Source combind guiley Currently 6 years of follow up with multiple adminsions: for expelation presuments. Surene opilipsy from 12/12 of age.
							Transcense aerebelter diameter 3 SD - man N varries N - SD - SD - man N varries N - M - M - M - M - M - M - M - M - M -			Puristust FOD -3rd certile, ACC, abnormal unlargement of the gauginosis emissence. Bilateral abnormality of opercularization,								
		Abnormal appearance of pos	tarios fosca, cyclic structure -				Francisco contribute diameter on 50th contile but artificially increased by dilated 4th ventricle displacing the carebollar likhteral of	th interhemispheric cyst. mild copocaphaly 10mm and	ACC, percietant Blake's goods, versuis and postine biometry	Purished FOD -Ash centles, ACC, absorbed onlingment of the gaughesis unincone. Educated absentably of opercubations, homopholic deprepties Ponders AP distance SDD below mean far GA and argumentic said pose on relail images. Purcinster distanced Blake's peach with version retains an aparticular relaced AP states distance. Consolidations of Endings highly neggestive of					ACC. Enlarged based gauglia with about nature limit internal superais. Edutated abN operatherisation and orteonized dyagotia. Businesses dyagotian with cased pose and argumentsy of vasted pose on axial images. Variety operated by previous Eduke's poach. Hypogylasia of ventus and unribullar humisphone. Funding			Saismon, severe developmental delay: 1 year and 10 months of follow up with multiple hospital admission
11		21+2 anchoold oper or litaku's pos	skit Psobable ACC. 22-2	Bate use 2 and 3 SD below mean	GE.	N	beariophores laterally. N (Innex at	rial diameter. 28-9	below 3rd percentile, extention moderately delayed for GA	6+0 toledinepathy.	8 N microactoy	ilrabons 6	81+2 yes	1	typical for tubulinopathy.	als I	Materiopygous for cl2MSG-A pathogonic variant in TURATA game	for aspiration procureous.
						Maked underconneclarization	This and kinked. Deep ventual pontine old to axial impace.		Walker Warbers shows tree, conductor in mornion affection									Document at 63 days of are. Principal diagnosus-
12		20/40 ACC: Pensionet Elakris pou	a. 24+1	2 SD below mean	BE, BC	Parietoccipital raisus and cinglolate raisus not ease. Sevuro lateral rantriculomogaly.	Dany ventral pontine chief on soil images. AP pontine diamete Transvarue combolite diameter 2 35D below mean for SD below mean for GA GA. ACC	32-2	Walter Washung phonotype, conholler hypoplasia afforting somis and homisphone, y-shaped kinking of this besinesses afti- homispheric paracelyses with abuset selectors. ACC, vantual postion cleft, hillstand savous VM.		la I N microstray	linabora.	80+1 yes	7	Cobblestons lineacophaly, ACC, kinked thin brainstom, conbollar hypophoia, ordayed dysmosphic based gauglia. Consistent with Walker Warburg phenotype.	ao PM I	Neuromecular disease gote pand analysis-multiple sequence variant detected but some considered distingly relevant. Tubulin gotes not included in texted panel	 Encocophaly, science, requiremy failure related to poor tree, namological feeding problems, diabetes incipidus, hypernationnia, capcie
		Missouduly biland summi	indonesia left Home side.			This parenclyms, underspecularization, moderately serves lateral contriculomogaly (13mm). Subopondymal accludos	This AP General of											
13		22+1 Huma, CSP and CC ort ide 22+1 to obvious varnis, brainstan	etifed, conbellum small with a small with flattuning of pone 22-6	35D below mean	BE	(13mm) . Subspendymal nodulos lining one frontal hora.	Thin, AP diameter of poor 3 SD below means for GA poor 3 SD below means for GA. ACC	ab.	aù.	a/a	h I N microscopy	TOP	23+3 80	*	ab	Facial dysmosphism, microcophaly, undordrockpad sukis, sunticulomogaly, ACC, hypoplanic conshellum 1	De novo pathogonic variant TURALA gene. Both parents are negative for the nutation, therefore variant suchnoided as pathogonic	-
14		Servare VM, 16mm artial dia material, consistant with olor 22+2 unabilium, CSP procent but	noter bilaterally, schogosic within ventricles, small thin CC noted 23+0	25D below mean	DE.	Underspecularization bilatarally. Bilataral VM (16mm and 17mm). This homophoric parandyma.	Varion height and transvaria corelector diameter 2 SD below the mean N Present a	and complete but aboresaily thin 25+0	Increased lexical VM now 15-Henra, hypogenic CC, failure of progression of primary exhaden since persions MR, continued workslike hypoplesia	ab a		TOP	15+2 ao		als.	severo brain macention, absent CC, fillend venericles 1	betavoypous various c.74G-T (p.C)s25Plu) in ocos 2 of TUBALA gone	eù.
							Vernis and transverses Not kinked. Press AP											
15		Bilatard corolon ventriculos 20+3 hasmorthago into the choroid	egaly 13 mm. Suspected places bilaterally 22=6	35D below mean	RE, RC	Savare vantriculomogaly and anderoparcularization.	Variation and transverserous Contribution Company of the Company o	ale	ala	6/8	. I N	Limbora famala 2	19+2 Y	6	Serven vanticulomogaly. Cobblismon Seamosphaly	als 6	nis henropyons variant in the TURATA gene v. 719C~G (p.Als200Gly)	Death at 4 months of age due to poor time / respirator failure / poor oradiorring
16.		19+3 Small corchellum	20-3	3 SD below mean	95	Stituted acclustely severe VM. This couled parentlyms. No significant operularization. This	care-boller dissenter 25D below ments ment ment ment ment ment ment ment ment	ab .	49	n's .	100	TOP	13+5 N	**	45	urbhinane Seuscophdy I	betweeppone various in the TUBAIA game a 790°-G (p.Ala/M6G)). However gone pand testing in 2014 for Spanes known to canon Walker - Washing syndroms was negative. Walker Stathing syndroms was negative. heterotygous various in the TUBAIA game a 790°-G (p.Ala/M6G); as for shilling. Gerelline measurisms in parent nespected due to	Elder chiling of 23
17		Moderate vustriculomogaly, 19+0 about CSP, micrographia	ippoplaric unubsilians, 21+1	2 SD below mean	26	No significant opercularization. Thin homipharic puresdown. Severe bilateral based VM	Reduced corebillar and varmin 2 SD below mean for districtor 2 SD below mean GA. Upward sotation of condulture	34-9	49	**		Limbon 3	15+3 N	**	44	1	as for obling. Geraline menalism in parent megocard due to excurrence of AD nomatic pathogenic metation in two oblings.	Naonatal death (day 1). Younger shiling of 2.2.
						No significant opercularization. Severe	Mild position kink. Position AP disasters: Descript AP disasters: Six Descript AP disasters: Six Descript AP disasters: Six Descript AP disasters: City and relation of conshellment CA. City and relation of conshellment City and relation of the conshellment C										Hateropygous minutes statistics TUBAIA. OMEM # Lineacophaly	
is is		21+4 Partnier focus abscensibly	Dandy Walker multi-creation? 24+4	2.5 SD below man for GA	10.	bnasil VM.	contin. positive delt. ACC				******	JOP	N N	43			A SE DROP ALL	
						Underopercularization. Underdoveloped primary and secondary caleston. Absorved	TCD and verses beight more								Troical factors of tabulicondry - Securconduly artifered hand			
19		als als	33+0	More than 2 SD below the mean for GA	ac	Underopercularization. Underdeveloped primary and successfury estimates. Aborated particularies of laminated appearance. Bilateral VM > 12mm.	Base 2SD < mean. Servere global carebollar hoposplasia	ab	83	n/a		Eirahora famala	40 Y	**	Typical funtures of tabulinopolty - Isoancaphaly celar god basal gaugha, absent autorier limb-of internal capeala, canabellar hypophoia	ab I	Betweeygous TUBAIA c 5977°-G (p Plu296Cys)	Epilopey
		Biland vanish	odanau right, severa left.															
30		Bilintaral vantriculomogaly m leypoplastic pone, abnormal l leypotalurium. 26+5	rain gyration and	25D races	as.	Absormal opercularization, dyagonia. Moderate right and severe left contriculomogaly.	Morphologically accessed. AP diameter of poor Transverse diameter 13th contile 10th contile for GA. No braineten kink. Present	ab.	43	6/8		TOP	N	**	49		Class Stpathogoich TUBBO variants: 1138C>Tap.Aug380Cys	
																	Oligophumin I sustation. dulation 3420 kp; NG 009960.1 a 242551 245371, Dulation of green 31 a 14 8 NA money.	
21			9 - 0	50th contile	ar.	N apart from mild bilatoral VM	N N		60			lirabora mala	v	2 years	Perioanticular nodular hataontopia. Persicunt maga cictuma magna. Mesial temporal sularusis.		OSpophanini I mutation. dulation 1820 bp. NG. 000960.1 g.24251; 245371. Dulation of uton 13 e 14 ENA mass. (Nd. 00247; 2.1185; 13914b) familial mutation and promutative step produce (Np. 0002381; p.18160 6*21); Same-dulation found in one X chromosome of the mether.	Fabrile soiranos. Mild convolves/openental delay.
						Bilataral abnormal operathrization			Subbetantial reduction in situ of GFs compand with prior examination but cell abornably large on the right, bilanced found subspendy and pseudocyces, CC present but thinted. Abournal operation into partials. Substatus has progressed appropriate for go periation.						Eight frontal polymicogyria, abountal orientation of Sylvian		Blood and chin microsumy - normal. Promuted biopey of filtachinid depigemented chin hoises (dark - skinned infam) falled to demonstrate enjourney matrices on W.S.	Solvense including infantle spacese, visual impairmen
EEGINB 22		22+3 Uniktural VM 12mm, difficu	by visualising CSP. 25+1	20th contile	BG, BC	and periophian polymicrogenia.	N N	22-1	for gentalon.	n/a	0 000	Sirabora	15+6 yes	59.	faccore, bilancial cubapondynial pseudocycte.	ab I	explanatory materiou on WTS.	developmental delay carebral palety
ALCC = supereion of the corpus califorare ACC = supereion of the corpus califorare AF = arteroposterior CSF = cassum septum pelludicum CC = corpus califorare GA = gostational age N = Normal																		
U. is corpus casouem GA = gestational age N = Normal n/a = not available or not applicable tp = not performed																		
to a me avateure o me appearante pa and performed TSC a tuberous sciencis complex VM = ventriculoringuly WS without moure respective																		
MES = whole exame sequencing																		
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