

Table 1
Clinical features of twelve novel PIGT-CDG cases and the clinical features in the fourteen previously published PIGT-CDG cases.

Physiologic System	Clinical	Kvarnung et al. 2013 [10]	Nakashima et al. 2014 [28]	Lam et al. 2015 [25]	Skauli et al. 2016 [27]	Pagnamenta 2017 [30]	Kohashi 2017 [26]	Patient 1	Patient 2	Patient 3	Patient 4	Patient 5	Patient 6	Patient 7	Patient 8	Patient 9	Patient 10	Patient 11	Patient 12	Patient 13	Patient 14
Sex / age at investigation		Turkish family. 4 affected females. Ages 1-3 years	Japanese female. 12-years-old	Caucasian mother and an African American father Patient 1: A 7-years-old female Patient 2: A 6-years-old male	Patient 1: African male, 9-years-old Patient 2: African male, 7-years-old	Patient 1: Caucasian female, age unknown Patient 2: Afghanistani male, age unknown	Japanese male, 11 months-old	Danish female, 7-years-old (family 1)	Danish male, deceased at 11 months of age (family 1)	Asian female, deceased at 6 months of age (family 2)	Polish female, 5-years-old (family 3)	Polish female, 9-years-old (family 4)	Polish female, 4-years-old (family 4)	Somalian male, 11-years-old (family 5)	Somalian male, 22-months-old (family 5)	Female (family 6)	Male, deceased at 26 months of age (family 6)	Polish female / 4-years-old (family 7)	Pakistani female, 2-years-old (family 8)	Bangladeshi female, 6-months-old (family 9)-	Bangladeshi female, 4-months-old (family 9)
Genetic investigation		Whole exome sequencing	Whole exome sequencing	Whole exome sequencing	Whole exome sequencing	Whole exome sequencing	Whole exome sequencing	Whole exome sequencing	Whole exome sequencing	Targeted NGS panel	Whole exome sequencing	Whole exome sequencing	Sanger sequencing	Exome sequencing via the DDD study	Sanger sequencing	Exome sequencing via the DDD study	Targeted Sanger sequencing after result in patient 9	Exome sequencing via the DDD study	Targeted NGS panel	Targeted NGS panel after result in patient 10	Whole exome sequencing
Genetic result		c.547A>C; c.547A>C	c.250G>T; c.1342C>T	c.918dupC; c.1342C>T	c.1079G>T; c.1079G>T	Patient 1: c.1582G>A; c.1730dupC Patient 2: c.709G>C; c.709G>C	c.250G>T; c.1096G>T	c.1472T>A; 1484+2T>A	c.1472T>A; 1484+2T>A	c.709G>C; c.709G>C	c.494-2A, c.1582G>A	c.1582G>A; c.1582G>A	c.1582G>A; c.1582G>A	c.1079G>T; c.1079G>T	c.1079G>T; c.1079G>T	c.1079G>T; c.1079G>T	c.1097G>T; c.1097G>T	c.1724_1725insC; c.1582G>A	c.550G>A; c.550G>A	c.709G>C; c.709G>C	c.709G>C; c.709G>C
Neonatal	Gestational age	37–40 weeks	40 weeks	31 weeks	40 weeks	Unknown	Unknown	40 weeks	39 weeks	39 weeks	39 weeks	38 weeks	37 weeks	42 weeks	29 weeks	40 weeks	41 weeks	42 weeks	36w	40 weeks	39 weeks
	Birth weight	69–99th centile	50–90th centile	10th and 90th centile	50th centile	Unknown	Unknown	90th centile	50th centile	0.4th centile	>90th centile	>95th centile	>95th centile	95th centile	Unknown	50th centile	99th centile	91th centile	72th centile	<0.4th centile	0.4th centile
	Birth length	93–99th centile	50th centile	10-90th centile	50th centile	Unknown	Unknown	95th centile	90th centile	Not recorded	>97th centile	>95th centile	>75th centile	Not known	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown
	Birth head circumference	84–99th centile	50–90th centile	<10-75th centile	10th and >97.5th centile	Unknown	Unknown	>95th centile	50th centile	Not recorded	50th centile	>95th centile	>95th centile	69th centile	Unknown	<0.4th centile	>95th centile	Not recorded	Not recorded	<0.4th centile	<0.4th centile
	Other	No	Polyhy	1/2				No	No								Electiv				

			dramnos	Elevated AFP on 2nd trimester screen	No	Unknown	Cesarean section due to breech presentation			Forceps assisted delivery	No	No	No	No	Unknown	Cesarean section due to breech presentation	elective cesarean section	Vaginal bleeding at 6-8 weeks and 12 weeks of pregnancy	Born by assisted vaginal delivery	Forceps assisted delivery, Bilateral elbow contractures & distal finger contractures	Multiple bone fractures	
Dysmorphic features	Skull	Brachycephaly (4/4)	Unknown	Brachycephaly (2/2)	Unknown (2/2)	Unknown	Unknown	Brachycephaly	Brachycephaly	Unknown	Unknown	Brachycephaly	No	Sagittal ridge	No	Metopic and sagittal ridge	Not at birth Later. Scaphocephaly	Normal	Brachycephaly	Normal	Normal	
	High forehead	4/4	Unknown	2/2	Yes	Yes (patient 1)	Unknown	Yes	Yes	Unknown	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Unknown	Yes
	Bitemporal narrowing	4/4	Unknown	2/2	Yes	Yes (patient 1)	Unknown	Yes	Yes	Unknown	Yes	NO	Yes	Yes	No	No	No	Yes	No	No	No	No
	Telecanthus	Unknown	Unknown	Unknown	Unknown	Yes (patient 1)	Yes (and also epicanthus)	Yes	Yes	Unknown	Yes	Yes	Yes	No	No	Mild telecanthus	Mild telecanthus	No	Yes	Unknown	Unknown	
	Palpebral fissures	Straight (4/4)	Upslanting	Upslanting (2/2)	Straight (2/2)	Straight (patient 1)	Unknown	Straight, normal length	Straight, normal length	Unknown	Straight	Upslanting	Upslanting	Mildly downslanting	No	Straight, normal length	Normal	Straight, normal length	Normal	Normal	Upslanting	
	Eyebrows	Arched (3/4)	Unknown	Arched (2/2)	Arched (1/2)	Straight (patient 1)	Arched	Straight	Straight	Unknown	Straight	Arched	Arched	Straight	Unknown	Straight	Normal	Normal	Normal	Normal	Normal	
	Nose	Short, anteverted (4/4)	Short, anteverted	Short, anteverted (2/2)	Short, anteverted (2/2)	Short with broad nasal tip (patient 1)	Short, anteverted	Short, anteverted	Short, anteverted	Unknown	Small	Short, anteverted	Short, anteverted	Short, anteverted	Unknown	Unknown	Normal	Broad nasal tip	Short anteverted	Normal	Normal	
	Nasal bridge	Depressed (4/4)	Depressed	Depressed (2/2)	Depressed (2/2)	Unknown	Depressed	Depressed	Depressed	Unknown	Depressed	Depressed	Depressed	Broad and depressed	Unknown	Depressed	Mildly depressed	Normal	Broad	Normal	Normal	
	Malar flattening	Yes (4/4)	Yes	Yes (2/2)	Yes (2/2)	Unknown	Unknown	Yes	Yes	Unknown	Yes	Yes	Yes	No	No	Yes	No	No	Yes	No	No	
	Philtrum	Long and distinct (4/4)	Unknown	Long (2/2)	Short (2/2)	Short (patient 1)	Long	Long and distinct	Long and distinct	Unknown	Normal	Long and distinct	Long and distinct	Normal	Normal	Short	Normal	Distinct	Distinct	Normal	Normal	
	Cupid bow lip	Yes (4/4)	Unknown	Yes (2/2)	Unknown	Unknown	Unknown (tent)	Yes	Yes	Unknown	Yes	Yes	Yes	No	No	Yes	Yes	Yes	Yes	Normal	Unknown	

							lip)																
	Mouth	Unknown	Downturned corners of mouth	2/2 Downturned corners of mouth	Tented upper lip (2/2)	Wide mouth (patient 1)	Unknown	Tented upper lip, microstomia	Tented upper lip, microstomia	Unknown	Tented upper lip	Prominent and everted lower lip	Prominent and everted lower lip	Tented upper lip Downturned corners	Tented upper lip	Tented upper lip	Tented upper lip	Unknown	Tented upper lip	Unknown	Unknown		
	High arched palate	Unknown	Yes	Yes (2/2)	Yes (2/2)	Unknown	Yes	Yes	No	Unknown	Not recorded	Yes	Yes	Yes	Yes	Unknown	Unknown	No	Yes	Yes	Unknown		
	Teeth abnormalities	Yes (4/4)	No	Unknown	Unknown	Unknown	Unknown	Yes	Yes	Unknown	Unknown	No	No	Yes	Unknown	No	Gum hypertrophy	No	Yes	Unknown	Unknown		
	Micrognathia	Yes (4/4)	Yes	No	No	No	Yes	Yes	Yes	Unknown	No	No	No	No	No	No	No	No	No	No	No		
	Ears	Unknown	Low set	Large ears (2/2)	Low set and (1/2)	Low set (patient 1)	Unknown	Low set and uplifted earlobes	Normal	Unknown	Normal	Normal	Normal	Low set	No	Normal	Normal	Normal	Normal	Normal	Normal	Low set	
	Hair	Hypotrichosis Thin, high frontal hairline.	Ingibilleder	Hypotrichosis Thin, high frontal hairline.	Hypotrichosis Thin, high frontal hairline. (2/2)	Unknown	Unknown	Hypotrichosis Thin, high frontal hairline. Hypertrichosis	Hypotrichosis Thin, high frontal hairline.	Unknown	Hypotrichosis, high frontal hairline	Hypotrichosis, Thin, high frontal hairline.	Hypotrichosis, Thin, high frontal hairline.	Hypotrichosis. Thin frontal hairline.	Hypotrichosis. Thin frontal hairline.	Unknown	Unknown	Unknown	Hypotrichosis. Thin frontal hairline.	Unknown	Unknown		
	Body proportions	Short arms (4/4)	Unknown	Normal (2/2)	Normal	Unknown	Unknown	Normal	Normal	Unknown	Normal	Relative macrocephaly	Relative macrocephaly	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal		
Neurological	Intellectual disability	Profound (4/4)	Profound	Profound (2/2)	Profound (2/2)	Profound (2/2)	Profound	Profound	Profound	Profound	Severe	Severe	Severe	Profound	Profound	Profound	Profound	Severe	Profound	Profound	Profound		
	Seizure onset	12-18 months of age	4 months of life	5 months of life	12 months of age	Patient 1: 12 months of age Patient 2 Neonatal onset	2 months of life	1th day of life	1th day of life	2 weeks of life	18 months of age	11 months of age	8 months of age	11 months of age	14 months of age	10 months of age	12 months of age	12 months of age	5 months	1st day of life	1th day of life		
	Epileptic seizures	Myoclonic (2/4). Generalized tonic clonic (1/4). Absence+head jerks &	Myoclonic, tonic with apnea that can generalize	Myoclonic, tonic, and tonic clonic that occasionally generalize (2/2)	Myoclonic, tonic, and tonic clonic that occasionally generalize (2/2)	Generalized tonic clonic (2/2)	Myoclonic, tonic with apnea that can generalize	Tonic and myoclonic with apnea. Subtle focal seizures. Recurrent CSE	Tonic and myoclonic with apnea. Subtle focal seizures. Recurrent CSE	Myoclonic and subtle focal (febrile / upper respiratory infection)	Tonic-clonic and generalized	Tonic-clonic and generalized	Tonic and myoclonic occasionally generalized with apnea	Generalized tonic clonic and atonic seizures	Myoclonic and focal seizures	Myoclonic and subtle focal seizures. Febrile seizures.	Myoclonic and focal seizures, Some generalized. Febrile seizure	Generalized tonic clonic seizures	Generalized tonic and myoclonic. Focal myoclonic seizures. Febrile seizures.	Myoclonic jerks.	Generalized tonic seizures.		

		blinking (1/4)														s.					
EEG features	Multifocal epileptiform abnormalities (3/4)	Multifocal epileptiform abnormalities (2/2)	Multifocal epileptiform abnormalities	(2 wks) bilateral burst of SW (4 yrs) BG slowing, focal FT slowing and SW	(2 wks) bilateral burst of SW (6 mo) BG slowing, focal posterior slowing and SW	(2 wks) burst suppression, myoclonic jerks, focal sz (right and left T)	(4 yrs) slowing and (poly)SW in the left FT region	(3 and 7 yrs) BG slowing	(18 mo) BG slowing, sporadic SW in the left FT region or F bilat	(22 mo) bilateral bursts of SW	NA	(10 mo) normal	(12 mo) bilateral fast activity (f.u) Multifocal epileptiform abnormalities	(12 mo) BG slowing,	(6 mo) BG slowing, posterior epileptiform abnormalities	(neonatal) burst suppression pattern	(age unknown) BG slowing and burst suppression pattern				
Seizure outcome	Intractable	Intractable	Intractable	Intractable	Intractable	Patient 1: Favorable (seizure-free with AED). Patient 2 Intractable	Intractable	Intractable	Intractable	Intractable	Favorable	Favorable	Favorable	Intractable on AEDs but almost seizure-free on ketogenic diet	Intractable	Intractable	Intractable	Favorable	Intractable	Intractable	Intractable
Epileptic encephalopathy or developmental delay with epilepsy	Epileptic encephalopathy	Patient 1: Developmental delay with epilepsy Patient 2 Epileptic encephalopathy	Epileptic encephalopathy	Epileptic encephalopathy	Epileptic encephalopathy	Epileptic encephalopathy	Developmental delay with epilepsy	Developmental delay with epilepsy	Developmental delay with epilepsy	Epileptic encephalopathy	Epileptic encephalopathy	Epileptic encephalopathy	Epileptic encephalopathy	Developmental delay with epilepsy	Epileptic encephalopathy	Epileptic encephalopathy	Epileptic encephalopathy				
Hypotonia	Yes (4/4)	Yes	Yes (2/2)	Yes (2/2)	Yes (2/2)	Yes (2/2)	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes but with brisk reflexes	Yes but with brisk reflexes	Yes	Yes	Yes	Yes
Was an MRI available	Yes (4/4)	Yes	Yes (2/2)	Yes (2/2)	Yes (2/2)	Yes (2/2)	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Brain imaging — Global cerebral and cerebellar atrophy with <u>predominant</u> vermis and cerebellar hemisphere	Yes (3/4)	Yes	Yes (2/2)	Yes (2/2)	Yes (2/2)	Progressive isolated cerebellar atrophy affecting vermis and cerebellar hemispheres (patient 1)	Progressive isolated cerebellar atrophy affecting vermis and cerebellar hemispheres	No	No	No	Yes	Yes	No	No (only slight asymmetry of the hippocampus)	No (only mild ventricular dilatation)	Yes	No	Progressive isolated cerebellar atrophy affecting vermis and hemispheres	Global cerebral and cerebellar atrophy.	Unknown	No

	atrophy																	(at 4 years of age).			
	Brain imaging — Possible neuronal migration defect	2/4	Unknown	0/2	Unknown	Unknown	Unknown	No	No	No	No	No	No	No	No	No	No	No	Unknown	Normal	Unknown
	CSF (glucose, protein, cell count, oligoclonal bands)	Unknown	Unknown	Normal (2/2)	Yes (2/2)	Unknown	Unknown	High protein count	Unknown	Normal	Unknown	Unknown	Unknown	Normal	Unknown	Normal	Unknown	Normal	Normal	Normal	Not performed
	CSF albumin & albumin quotient	Unknown	Unknown	Low (2/2)	Unknown	Unknown	Unknown	High	Unknown	Normal	Unknown	Unknown	Unknown	Unknown	Unknown	Normal	Normal	Unknown	Unknown	Normal	Unknown
	Peripheral nerve conduction studies	Unknown	Unknown	Normal (2/2)	Unknown	Unknown	Unknown	Not performed	Not performed	Not performed	Unknown	Not performed	Not performed	Normal	Unknown	Not performed	EMG and nerve conduction did not show any evidence of neuromuscular problem but neurogenic change in bulbar musculature	Not performed	Normal	Not performed	Not performed
Ophthalmologic	Cortical visual impairment	Yes (4/4)	Yes	Yes (2/2)	Yes (2/2)	Unknown	Unknown	Yes	Yes	No but ophthalmological examination was done once at three weeks of age.	Unknown	Yes	Yes	No	Yes	Unknown	No	No	Yes	No	No
	Strabismus, nystagmus	Yes (4/4)	Yes	Yes (2/2)	Yes (2/2)	Unknown	Unknown	Yes	Yes	No	No	Yes	Yes	No	Yes	Yes	Anterior convergent Down beat	No	Yes	Unknown	Unknown

																	nystagmus					
	Hyperopia	Yes (4/4)	Unknown	1/2	Unknown	Unknown	Unknown	No	Unknown	No	Yes	Yes	Yes	No	No	No	Yes	No	Unknown	Unknown	Unknown	
	Myopia	0/4	Unknown	1/2	Unknown	Unknown	Unknown	Yes	Unknown	No	No	No	No	No	No	No	No	No	Unknown	Unknown	Unknown	
	Other	Down gaze palsy (1/4)	Unknown	Astigmatism (1/2) Posterior staphylocoma (1/2)	Astigmatism (2/2)	Oculomotor apraxia (patient 1). Nystagmus and optic atrophy (patient 2)	Unknown			No	No	Astigmatism	Astigmatism	No	No	No	Deep optic discs	Vertical oculomotor apraxia, previous episodes of tonic upgaze deviation	No	Unknown	Unknown	
Audiologic	Hearing loss	0/4	Unknown	2/2	Unknown	Unknown	Unknown	No	No	No	Unknown	No	No	No	Normal	Unknown	Unknown	No	Normal	Normal	Unknown	
Cardiologic	Patent ductus arteriosus	1/4	Yes	0/2	No	Unknown	No	No	No	No	No	No	No	PFO with an atrial septal aneurysm	No	Small PDA	Unknown	No	No	No	No	
	Restrictive cardiomyopathy	1/4	No	0/2	No	Unknown	No	No	No	No	No	No	No	No	No	No	Unknown	No	No	No	No	
	Increased atrial load	1/4	No	1/2	No	Unknown	No	No	No	No	No	Unknown	Unknown	No	No	No	Recurrent bradycardia and atrial ectopics noted on ECG- no cardiac echo performed	No	No	No	No	
	Atrial Septal defect	0/4	No	1/2	No	Unknown	No	No	No	No	No	No	No	No	No	Unknown	Unknown	No	No	No	No	
	Borderline long QT	Unknown	Unknown	1/2	Unknown	Unknown	No	No	Unknown	Unknown	Unknown	No	No	No	No	Unknown	No	Unknown	No	No	Unknown	
Respiratory		Atypical lung lobulation	Unknown	Mixed central and	Unknown	Unknown	Central apnea	Central sleep apnea	Central sleep apnea	None	Unknown	No	No	None	Mild obstructive sleep apnea	Obstructive sleep apnea	apnea	None	Central sleep apnea	No	No	

		(1/4)		obstructive sleep apnea (1/2)																	
Gastro-intestinal	Obesity	No	Unknown	Yes (2/2)	Unknown	Unknown		No	No	No	No	Unknown	No								
	Gastroesophageal reflux	Unknown	Unknown	Yes (2/2)	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	No	No	Yes	Yes	No	No	Unknown	Yes	Yes	Unknown
	Aspiration risk + secondary G-tube dependence	Unknown	Unknown	Yes (2/2)	Unknown	Unknown	Yes	Yes	Yes	Yes	Unknown	Unknown	Unknown	No	Impaired swallow	Unknown	Yes	No	Yes	Unknown	Yes
	Hypertriglyceridemia	Unknown	Unknown	1/2	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Yes	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	No	Unknown
Urologic/renal	Nephrocalcinosis	Yes (4/4)	No (but + urolithiasis)	No	No	Nephrolithiasis (patient 1)	Unknown	No	No	No	Unknown	No	No	No	Unknown	No	Unknown	Left ureteric stone	No	Unknown	No
	Urine calcium	High	Normal	Normal	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	No	Unknown	No	Unknown	Normal	Unknown	No	Unknown
	Ureteral dilation	Yes (3/4)	Yes	0/2	No	Unknown	Unknown	No	Unknown	No	Not known	No	Yes	No	Unknown						
	Renal Cysts and dysplasia	¼	No	0/2	No	Unknown	Unknown	No	Unknown	No	Unknown	No	No	Unknown	No						
Musculoskeletal	Premature loss of teeth	2/4	No	0/2	Unknown	Unknown	Unknown	No	Unknown	Unknown	No	No	No	No	Unknown	No	No	No	No	No	Unknown
	Slender long bones	4/4	Unknown	2/2	No (but pictures not shown)	Unknown	Unknown	Yes	Yes	Unknown	Unknown	No	No	Unknown	Unknown	Unknown	Not known	No	Unknown	No	Yes
	Scoliosis	2/4	Yes	2/2	No	Unknown	Unknown	No	No	No	No	Yes	No	No	No	Unknown	No	No	No	No	No
	Craniosynostosis	2/4	No	Unknown	No	Unknown	Unknown	Unknown	Unknown	Unknown	No	No	No	Unknown	No	Metopic and saggital ridge		No	Unknown	No	No
	Short arms	4/4	Unknown	0/2	No	Unknown	Unknown	No	No	No	No	Unknown	Unknown	No	No	Unknown	No	No	No	No	No
	Pectus excavatum	1/4	Unknown	¼	No	Unknown	Unknown	No	Unknown	No	No	Yes	Unknown	No							

	m		wn																			
	Joint hypermobility	Unknown	Unknown	2/2	Unknown	Unknown	Unknown	Yes	Yes	No	No	Yes	Yes	No	No	Unknown	Unknown	Unknown	Yes	No	Unknown	
	Electromyography	Unknown	Unknown	Reduced muscle bulk, but normal motor units	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Neurogenic change in the bulbar musculature otherwise normal EMG and NC	Unknown	No	Unknown	No	
Endocrine	Bone age	Delayed (4/4)	Unknown	Advanced (1/2), Normal (1/2)	Normal (2/2)	Unknown	Unknown	Normal	Unknown	Unknown	Unknown	Normal	Unknown	Unknown	Unknown	Unknown	Not known	Unknown	Unknown	Unknown	Unknown	
	Osteopenia	4/4	Yes	2/2	No (2/2)	Unknown	Unknown	Yes	Yes	Unknown	Unknown	Unknown	Unknown	Unknown	No	Unknown	Not known	Unknown	Unknown	Normal	Yes	
	Plasma alkaline phosphatase	Low	Low	Normal	Normal	Normal (patient 1) Low (patient 2)	Low	Normal	Normal	Unknown	Normal	Normal										
	Plasma calcium	High/high normal	Normal	Normal	Normal	Unknown	Not specified but laboratory tests were described as normal	Normal	Normal	Normal	Unknown	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Unknown	Normal	Normal	
	Plasma phosphate	Normal	Unknown	Normal	Normal	Unknown	Not specified but laboratory tests were described as normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	No	Unknown	Normal	Normal	Unknown	Unknown	Normal
	Parathyroid hormone values	Low	Unknown	Normal	Unknown	Unknown	Not specified but laboratory tests were described as normal	Normal	Normal	Normal	Unknown	Normal	Unknown	Normal	Unknown							

	Thyroid function tests	Normal	Unknown	Normal	Normal	Unknown	Not specified but laboratory tests were described as normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Normal	Unknown	Not done at GOSH	Normal	Unknown	Unknown	Normal
	Precocious puberty	0/4	Unknown	½	Unknown	Unknown	Not specified but laboratory tests were described as normal	Unknown	Unknown	Unknown	Not applicable	Unknown	Unknown	No	No	Unknown	Unknown	No	Unknown	Unknown	Unknown
Immunologic	IgA and IgM deficiency	Unknown	Unknown	½	Unknown	Unknown	Not specified but laboratory tests were described as normal	IgA and IgM deficiency	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Normal	Unknown	Unknown	IgA low, IgM lower limit of normal	No	Unknown	Unknown
	Other immune deficiencies	Unknown	Unknown	0/2	Unknown	Unknown	Not specified but laboratory tests were described as normal	No	No	Unknown	Unknown	No	No	Unknown	Pneumococcal meningitis and septicemia	Unknown	Bronchitis and chest infections	Unknown	Unknown	Unknown	Unknown
Hematologic	Hyper/hypo coagulability	Unknown	Unknown	0/2	Unknown	Unknown	Not specified but laboratory tests were described as normal	Normal	Normal	Unknown	Unknown	Normal	Normal	Unknown	Unknown	Unknown	No	Unknown	Unknown	Unknown	Unknown
	Factor excess or deficiency	Unknown	Unknown	Slightly decreased Factors V & XI (1/2) Mildly increased Factor VIII (1/2)	Unknown	Unknown	Not specified but laboratory tests were described as normal	Unknown	No	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown	Unknown

Abbreviations:

AFP: Alpha-fetoprotein
BG: background
CDG: Congenital disorders of glycosylation
CSE: Convulsive status epilepticus
DDD: Deciphering Developmental Disorders
EMG: Electromyography
F: frontal
NGS: Next generation sequencing
PFO: Persistent foramen ovale
PIGT: Phosphatidylinositol-glycan class T
SW: spike and waves
Sz: seizures
T: temporal