Supplementary Table 1: Overview of the reported variations of GPI-APs and sALP in known IGDs

	PIGA (1-2) n = 3 2 + 1	**PIGA** (3) n = 3	**PIGA** (4) n = 5	**PIGA** (5) n = 5	**PIGA** (6) n = 1	**PIGA** (7) n = 1	**PIGA** (8) n = 8		**PIGB** n = 14		**PIGC** n = 3		**PIGG** (1) n = 5	**PIGG** (2) n= 2
Ref	(1, 2)	(3, 4)	(5)	(4, 6)	(7)	(8)	(9)		(10)		(11)		(12)	(13)
CD14		**N** (Mon-1/1) n/a = 2									↓ (Gr-2/2) n/a = 1			
CD15														
CD16		↓ (Gr-1/1) n/a = 2		↓ (Gr-5/5)		↓ (RBCs) **N** (Leucos)			↓ (Gr-4/4) n/a = 10		↓ (Gr-2/2) n/a = 1		**N** (Gr-1/5) n/a = 4	
CD18														
CD24		↓ (Gr-1/1) n/a = 2							↓ (Gr-2/2) n/a = 12				**N** (Gr-1/5) n/a = 4	
CD33														
CD48													**N** (LCls-3/5) n/a = 2	
CD49														
CD55/DAF									**N** (Gr-2/3) ↓ (Gr-1/3) n/a = 11		↓ (Gr-2/2) n/a = 1		**N** (Gr-1/5) n/a = 4 **N** (LCls-3/5) n/a = 2	↓ (Fb-2/2) **N** (Gr-2/2)
CD59		**N** (RBCs-1/1) n/a = 2	**N** (LCLs-1/1) n/a = 4			**N** (RBCs)			↓ (Gr-3/3) n/a = 11 **N** (LCLs-2/2) n/a = 12		**N** (Gr-2/2) n/a = 1		**N** (Gr-1/5) n/a = 4 **N** (LCls-3/5) n/a = 2	↓ (Fb-2/2) **N** (Gr-2/2)
CD66b														
CD66c														
CD73									↓ (Fb-2/2) n/a = 12					↓ (Fb-2/2) **N** (Gr-2/2)
CD87														
CD90														↓ (Fb-2/2) **N** (Gr-2/2)
CD109					↓ (Fb)				**N** (Fb-1/2) ↓ (Fb-1/2) n/a = 12					
CD157														
FLAER		↓ (Gr-1/1) n/a = 2 **N** (Mon-1/1) n/a = 2		↓ (Gr-5/5)		**N** (Leucos)			**N** (Gr-3/4) ↓ (Gr-1/4) n/a = 10		**N** (Gr-1/2) ↓ (Gr-1/2) n/a = 1		**N** (Gr-1/5) n/a = 4	**N** (Fb,Gr-2/2)
T5 Ab														
sALP	↑ (3/3)	n/a	n/a	↑ (2/4) **N** (2/4) n/a = 1	↑	**N**	↑ (2/3) **N** (1/3) n/a = 5		↑ (8/9) **N** (1/9) n/a = 5		**N** (3/3)		**N** (3/3) n/a = 2	**N** (1/1) n/a = 1

sALP: serum alkaline phosphatase**; ↑**: elevated; **↓:** decreased**; n/a**: not available**; Gr**: granulocytes; **Mon**: monocytes; **MNC**: mononuclear cells; **RBCs**: red blood cells; **LCLs**: lymphoblastoid cell lines; **Leucos**: leucocytes; **Ly**: lymphocytes; **Fb**: fibroblasts; **BCs**: blood cells; **DAF**: decay accelerating factor; **R/PILC**: resistant to phosphatidylinositol-specific phospholipase C.

Supplementary Table: Overview of the reported variations of GPI-APs and sALP in known IGDs (continued)

	PIGH (1) n = 2	**PIGH** (2) n = 1		**PIGK** n = 12		**PIGL** (1) n = 6	**PIGL** (2) n = 1	**PIGL** (3) n = 1	**PIGL** (4) n = 3	**PIGL** (5) n = 1	**PIGL** (6) n = 2	**PIGL** (7) n = 2
Ref	(14)	(15)		(16)		(17, 18)	(19)	(18, 20)	(21)	(18)	(22)	(23)
CD14									↓ (Mon-3/3)			
CD15												
CD16	**N** (Gr-2/2)	↓ (Gr)		↓ (Gr-6/6) n/a = 6			↓ (Gr)		↓ (Gr-3/3)			↓ (Gr, Mon-2/2)
CD18												
CD24							↓ (Gr)		↓ (Gr-3/3)			↓ (Gr, Mon-2/2)
CD33												
CD48												
CD49												
CD55/DAF		↓ (Gr)					↓ (Gr)		↑ (Gr-3/3)			
CD59		**N** (Gr)							**N** (Gr-2/3) ↓ (Gr-1/3)			
CD66b												
CD66c												
CD73				↓ (Fb-2/2) n/a = 10								
CD87												
CD90												
CD109				↓ (Fb-2/2) n/a = 10								
CD157												
FLAER		**N** (Gr)		↓ (Gr-5/6) **N** (Gr-1/6) n/a = 6 ↓ (Fb-2/2) n/a = 10		↓ (Fb-1/1) n/a = 5 ↓ (LCLs-1/1) n/a = 5	↓ (Gr)		↓ (Gr-3/3)			↓ (Gr, Mon-2/2) ↓ (Ly-1/2) n/a = 1
T5 Ab												
sALP	**N** (2/2)	**N**		**N** (8/10) ↓ (2/10) n/a = 2		↑1/1 n/a = 5	↑	↑	**N** (3/3)	**+/-↑**	↑ (2/2)	n/a

sALP: serum alkaline phosphatase**; ↑**: elevated; **↓:** decreased**; n/a**: not available**; Gr**: granulocytes; **Mon**: monocytes; **MNC**: mononuclear cells; **RBCs**: red blood cells; **LCLs**: lymphoblastoid cell lines; **Leucos**: leucocytes; **Ly**: lymphocytes; **Fb**: fibroblasts; **BCs**: blood cells; **DAF**: decay accelerating factor; **R/PILC**: resistant to phosphatidylinositol-specific phospholipase C.

Supplementary Table: Overview of the reported variations of GPI-APs and sALP in known IGDs (continued)

	PIGM (1) n = 3	**PIGM** (2) n = 3		**PIGN** (1) n = 7	**PIGN** (2) n = 2	**PIGN** (3) n = 1	**PIGN** (4) n = 1	**PIGN** (5) n = 6	**PIGN** (6) n = 7
Ref	(24, 42)	(25)		(4, 26)	(4, 27)	(28)	(4, 29)	(30)	(9)
CD14		↓ (Gr, Mon, RBCs-3/3)							
CD15									
CD16					↓ (Gr-2/2)	N	**N** (Gr)		↓ (Gr-1/1) n/a = 6
CD18						↓ (Gr)			
CD24	↓ (Gr-2/2) n/a = 1	↓ (Gr, Mon, RBCs-3/3)			↓ (Gr-2/2)	↓ (Gr)	↓ (Gr)		↓ (Gr-1/1) n/a = 6
CD33									
CD48					**N** (LCLs-2/2)				
CD49									
CD55/DAF				↓ (Fb-2/2) n/a = 5	↓ (Gr-2/2) **N** (LCLs-2/2)				
CD59	↓ (Gr-2/2) n/a = 1 ↓ (Fb-1/1) n/a = 2 **~N** (RBCs, Plat-2/2) n/a = 1	↓ (Gr, Mon, RBCs-3/3) ↓ (Fb-1/1) n/a = 2		↓ (Fb-2/2) n/a = 5	**N** (LCLs-2/2)				↓ (Gr-1/1) n/a = 6
CD66b									
CD66c									
CD73				↓ (Fb-2/2) n/a = 5					
CD87		↓ (Fb-1/1) n/a = 2							
CD90									
CD109									
CD157									
FLAER	↓ (Gr-2/2) n/a = 1 ↓ (Fb-1/1) n/a = 2			↓ (Fb-2/2) n/a = 5	↓ (Gr-2/2) **N** (LCLs-2/2)	↓ (Gr)	↓ (Gr)		↓ (Gr-1/1) n/a = 6
T5 Ab									
sALP	**N** (3/3)	**N** (2/3) ~ ↑ (1/3)		n/a	**N** (1/1) n/a = 1	n/a	**N**	**N** (1/1) n/a = 5	**N** (4/5) ↑ (1/5) n/a = 2

sALP: serum alkaline phosphatase**;** ↑: elevated; **↓:** decreased**; n/a**: not available**; Gr**: granulocytes; **Mon**: monocytes; **MNC**: mononuclear cells; **RBCs**: red blood cells; **LCLs**: lymphoblastoid cell lines; **Leucos**: leucocytes; **Ly**: lymphocytes; **Fb**: fibroblasts; **BCs**: blood cells; **DAF**: decay accelerating factor; **R/PILC**: resistant to phosphatidylinositol-specific phospholipase C.

Supplementary Table: Overview of the reported variations of GPI-APs and sALP in known IGDs (continued)

	PIGO (1) n = 3	**PIGO** (2) n = 2	**PIGO** (3) n = 1	**PIGO** (4) n = 1	**PIGO** (5) n = 6	**PIGO** (6) n = 1	**PIGO** (7) n = 2	**PIGO** (8) n = 1		**PIGP** (1) n = 2	**PIGP** (2) n = 1	**PIGP** (3) n = 4		**PIGQ** (1) n = 1	**PIGQ** (2) n = 1
Ref	(31)	(32)	(33)	(20)	(34)	(35)	(36)	(37)		(38)	(39)	(40)		(41)	(42, 43)
CD14															
CD15															
CD16		**↓** (Gr) n/a = 1			**↓** (Gr-4/4) n/a = 2		**N** (Gr-1/2) n/a = 1			**↓** (Gr-1/1) n/a = 1		**↓** (Gr-2/2) n/a = 2			
CD18															
CD24		**↓** (Gr) n/a = 1			**↓** (Gr-4/4) n/a = 2		**N** (Gr-1/2) n/a = 1					**↓** (Gr-1/2) **N** (Gr-1/2) n/a = 2			
CD33															
CD48															
CD49															
CD55/DAF										**↓** (Gr-1/1) n/a = 1 **↓** (Ly-1/1) n/a = 1		**N** (Gr-1/1) n/a = 3			
CD59							**↓** (Gr-1/2) n/a = 1			**↓** (Gr-1/1) n/a = 1 **↓** (Ly-1/1) n/a = 1	**↓** (Gr)	**N** (Gr-1/1) n/a = 3			
CD66b															
CD66c															
CD73															
CD87															
CD90															
CD109															
CD157											**↓** (Gr)				
FLAER		**N** (Gr) n/a = 1			**↓** (Gr-3/4) **N** (Gr-1/4) n/a = 2					**↓** (Gr-1/1) n/a = 1	**↓** (Gr)	**N** (Gr-1/2) n/a = 3			
T5 Ab															
sALP	**↑** (3/3)	**~↑** (1/1) n/a = 1	**~↑**	**+/-↑**	**↑** (6/6)	**↑**	**+/-↑** (2/2)	**↑**		**N** (2/2)	n/a	**N** (1/1) n/a = 3		**? N**	**↑**

sALP: serum alkaline phosphatase**; ↑**: elevated; **↓:** decreased**; n/a**: not available**; Gr**: granulocytes; **Mon**: monocytes; **MNC**: mononuclear cells; **RBCs**: red blood cells; **LCLs**: lymphoblastoid cell lines; **Leucos**: leucocytes; **Ly**: lymphocytes; **Fb**: fibroblasts; **BCs**: blood cells; **DAF**: decay accelerating factor; **R/PILC**: resistant to phosphatidylinositol-specific phospholipase C.

Supplementary Table: Overview of the reported variations of GPI-APs and sALP in known IGDs (continued)

	PIGS n = 6		**PIGT** (1) n = 4	**PIGT** (2) n = 2	**PIGT** (3) n = 1	**PIGT** (4) n = 1	**PIGT** (5) n = 1	**PIGT** (6) n = 2	**PIGT** (7) n = 1	**PIGT** (8) n = 13	**PIGT** (9) n = 2	**PIGT** (10) n = 2	**PIGT** (11) n = 3
Ref	(44)		(4, 45)	(20)	(46)	(47)	(4, 48)	(4, 49)	(50)	(51)	(4, 52)	(9)	(4)
CD14											↓ (Mon-1/1) n/a = 1		
CD15													
CD16	↓ (Gr-4/4) n/a = 2		↓ (Gr-3/3) n/a = 1	↓ (Gr-1/1) n/a = 1	↓ (Gr-1/1)	↓ (BCs)	↓ (Gr)	↓ (Gr-2/2)			↓ (Gr-1/1) n/a=1		↓ (Gr-1/1) n/a = 2
CD18													
CD24						↓ (BCs)	**N** (Gr)				↓ (Gr, LyB-1/1) n/a = 1		
CD33													
CD48											↓ (Mon-1/1) n/a = 1		
CD49											~↓ (Fb-2/2)		
CD55/DAF	↓ (Gr-2/4) n/a = 4							↑ (Gr-2/2)	↓ (RBCs, Gr)				↓ (Fb-2/3) **N** (Fb-1/3)
CD59	↓ (Gr-2/4) n/a = 4		↓ (MNC-2/2) n/a = 2			**N** (BCs)		↑ (Gr-2/2)	↓ (RBCs, Gr)				↓ (Fb-3/3)
CD66b								↓ (Gr-1/2) ↑ (Gr-1/2)					
CD66c													
CD73													↓ (Fb-3/3)
CD87													
CD90											~↓ (Fb-2/2)		
CD109													
CD157													
FLAER	↓ (Ly-4/4) n/a = 2		↓ (Gr-2/3) **N** (Gr-1/3) n/a = 1		**N** (Gr-1/1)	↓ (BCs)	↓ (Gr)	↓ (Gr-2/2)			↓ (leucos-1/1) n/a = 1 ↓ (Fb-2/2)		↓ (Fb-2/3) N (Fb-1/3) ↓ (Gr-1/1) n/a = 2
T5 Ab													
sALP	**N** (3/4) ↓ (1/4) n/a = 2		↓ (4/4)	**N** (1/2) ~↓ (1/2)	↓ (1/1)	**N**	↓	**N** (2/2)	n/a	**N** (12/12) n/a = 1	n/a	**N** (1/1) n/a = 1	↓ (?)

sALP: serum alkaline phosphatase**;** ↑: elevated; **↓:** decreased**; n/a**: not available**; Gr**: granulocytes; **Mon**: monocytes; **MNC**: mononuclear cells; **RBCs**: red blood cells; **LCLs**: lymphoblastoid cell lines; **Leucos**: leucocytes; **Ly**: lymphocytes; **Fb**: fibroblasts; **BCs**: blood cells; **DAF**: decay accelerating factor; **R/PILC**: resistant to phosphatidylinositol-specific phospholipase C.

Supplementary Table: Overview of the reported variations of GPI-APs and sALP in known IGDs (continued)

	PIGU n = 5		**PIGV** (1) n = 8	**PIGV** (2) n = 2	**PIGV** (3) n = 9	**PIGV** (4) n = 1	**PIGV** (5) n = 2		**PIGW** (1) n = 1	**PIGW** (2) n = 1	**PIGW** (3) n = 2	**PIGW** (4-5) n = 2 1+ 1		**PIGY** n=4
Ref	(53)		(4, 54-58)	(4, 58)	(59)	(33)	(60)		(61)	(62)	(63)	(64, 65)		(66)
CD14									↑ (Gr) ↓↓(Mon) **N** (Ly)					
CD15									↓ (Gr) ↑ (Mon) **N** (Ly)					
CD16	↓ (Gr-5/5)		↓ (Gr-3/3) n/a = 5						↓ (Gr) **N** (Mon) ↓ (Ly)	↓ (Gr)	↓ (Gr-2/2)			**N** (Gr-2/2) n/a = 2
CD18														
CD24	↓ (Gr-5/5)								↓ (Gr) ↑ (Mon) ↑ (Ly)	↓ (Gr)	↓ (Gr-2/2)			
CD33									↓ (Gr) ↑ (Mon) **N** (Ly)					
CD48														
CD49														
CD55/DAF	**N** (Gr-5/5)		↓ (Fb-2/2) n/a = 6	↓ (Fb-1/1) n/a = 1					↓ (Gr, Mon, Ly)					↓ (Fb-2/2) n/a = 2
CD59	**N** (Gr-5/5)		↓ (Fb-2/2) n/a = 6	↓ (Fb-1/1) n/a = 1					↓ (Gr) ↑ (Mon) ↓ (Ly)					↓ (Fb-2/2) n/a = 2
CD66b														
CD66c											↓ (Gr-2/2)			
CD73			↓ (Fb-2/2) n/a = 6	↓ (Fb-1/1) n/a = 1										
CD87														
CD90														
CD109														
CD157														
FLAER	↓ (Gr-5/5)		↓ (Gr-3/3) n/a = 5 ↓ (Fb-2/3) n/a = 6	↓ (Fb-1/1) n/a = 1					↓ (Gr, Mon, Ly)	↓ (Gr)				
T5 Ab	↑ (5/5)													
sALP	**N** (5/5)		↑ (8/8)	↑ (2/2)	↑ (9/9)	↑	↑ (1/1) n/a = 1		↑	↑	**N** (2/2)	↑ (2/2)		↑ (2/4) **N** (2/4)

sALP: serum alkaline phosphatase**; ↑**: elevated; **↓:** decreased**; n/a**: not available**; Gr**: granulocytes; **Mon**: monocytes; **MNC**: mononuclear cells; **RBCs**: red blood cells; **LCLs**: lymphoblastoid cell lines; **Leucos**: leucocytes; **Ly**: lymphocytes; **Fb**: fibroblasts; **BCs**: blood cells; **DAF**: decay accelerating factor; **R/PILC**: resistant to phosphatidylinositol-specific phospholipase C.

Supplementary Table: Overview of the reported variations of GPI-APs and sALP in known IGDs (continued)

	PGAP1 (1) n = 2	**PGAP1** (2) n = 1	**PGAP1** (3) n = 2		**PGAP2** (1) n = 7	**PGAP2** (2) n = 2	**PGAP2** (3) n = 1	**PGAP2** (4-6) n = 9 2 + 4 + 3		**PGAP3** (1) n = 5	**PGAP3** (2) n = 8	**PGAP3** (3-8) n = 31 10 + 2 + 1 + 2 + 14 + 2		**GPAA1** n = 10
Ref	(67)	(68)	(69)		(70)	(71)	(72)	(73-75)		(76)	(4, 77)	(78-83)		(84)
CD14														
CD15														
CD16										↓ (Gr, Mon-1/1) n/a = 4				↓ (Gr-3/5) n/a = 5
CD18														
CD24							**N** (Gr)			↓ (Gr, Mon-1/1) n/a = 4				~↓ (LCLs-5/5) n/a = 5
CD33														
CD48	**N** (LCLs-1/1) **R/PIPLC** n/a = 1	**N** (LCLs) **R/PIPLC**												
CD49														
CD55/DAF	**N** (LCLs-1/1) **R/PIPLC** n/a = 1	**N** (LCLs) **R/PIPLC**			**N** (LCLs-3/3) n/a = 4						↓ (Fib-3/3) n/a = 5			
CD59	**N** (LCLs-1/1) **R/PIPLC** n/a = 1	**N** (LCLs) **R/PIPLC**			**N** (LCLs-3/3) n/a = 4		**N** (RBCs)				↓ (Fib-3/3) n/a = 5			
CD66b														
CD66c														
CD73											↓ (Fib-3/3) n/a = 5			
CD87														
CD90														
CD109														
CD157							**N** (Mon)							
FLAER							**N** (Gr, Mon)			↓ (Gr, Mon-1/1) n/a = 4	↓ (Fib-3/3) n/a = 5			↓ (Gr-3/5) n/a = 5 ↓ (LCLs-5/5) n/a = 5
T5 Ab														
sALP	n/a*	n/a*	**N** (2/2)		↑ (2/2) n/a = 5	↑ (2/2)	↑	↑ (9/9)		↑ (5/5)	↑ (8/8)	↑ (31/31)		**N** (10/10)

sALP: serum alkaline phosphatase**;** ↑: elevated; **↓:** decreased**; n/a**: not available**; Gr**: granulocytes; **Mon**: monocytes; **MNC**: mononuclear cells; **RBCs**: red blood cells; **LCLs**: lymphoblastoid cell lines; **Leucos**: leucocytes; **Ly**: lymphocytes; **Fb**: fibroblasts; **BCs**: blood cells; **DAF**: decay accelerating factor; **R/PILC**: resistant to phosphatidylinositol-specific phospholipase C.
***** Theoretically normal. Mentioned as 'normal' in ref (42), which indicates 11 PGAP1 patients.

References of Supplementary Table

1. Johnston JJ, Gropman AL, Sapp JC, Teer JK, Martin JM, Liu CF, et al. The Phenotype of a Germline Mutation in PIGA: The Gene Somatically Mutated in Paroxysmal Nocturnal Hemoglobinuria. The American Journal of Human Genetics. 2012;90(2):295-300.
2. Crabben SN van der, Harakalova M, Brilstra EH, Berkestijn FMC van, Hofstede FC, Vught AJ van, et al. Expanding the spectrum of phenotypes associated with germline PIGA mutations: A child with developmental delay, accelerated linear growth, facial dysmorphisms, elevated alkaline phosphatase, and progressive CNS abnormalities. American Journal of Medical Genetics Part A. 2014;164(1):29-35.
3. Swoboda KJ, Margraf RL, Carey JC, Zhou H, Newcomb TM, Coonrod E, et al. A novel germline PIGA mutation in Ferro-Cerebro-Cutaneous syndrome: A neurodegenerative X-linked epileptic encephalopathy with systemic iron-overload. American Journal of Medical Genetics Part A. 2014;164(1):17-28.
4. Knaus A, Pantel JT, Pendziwiat M, Hajjir N, Zhao M, Hsieh T-C, et al. Characterization of glycosylphosphatidylinositol biosynthesis defects by clinical features, flow cytometry, and automated image analysis. Genome Med. 2018;10(1):1-13.
5. Belet S, Fieremans N, Yuan X, Van Esch H, Verbeeck J, Ye Z, et al. Early frameshift mutation in PIGA identified in a large XLID family without neonatal lethality. Hum Mutat. 2014;35(3):350-5.
6. PIGA mutations cause early-onset epileptic encephalopathies and distinctive features | Neurology [Internet]. https://n.neurology.org/content/82/18/1587.long
7. Tarailo-Graovac M, Sinclair G, Stockler-Ipsiroglu S, Van Allen M, Rozmus J, Shyr C, et al. The genotypic and phenotypic spectrum of PIGA deficiency. Orphanet J Rare Dis. 2015;10:23.
8. Kim YO, Yang JH, Park C, Kim SK, Kim M-K, Shin M-G, et al. A novel PIGA mutation in a family with X-linked, early-onset epileptic encephalopathy. Brain Dev. 2016;38(8):750-4.
9. Jiao X, Xue J, Gong P, Bao X, Wu Y, Zhang Y, et al. Analyzing clinical and genetic characteristics of a cohort with multiple congenital anomalies-hypotonia-seizures syndrome (MCAHS). Orphanet J Rare Dis. 2020;15(1):78.
10. Murakami Y, Nguyen TTM, Baratang N, Raju PK, Knaus A, Ellard S, et al. Mutations in PIGB Cause an Inherited GPI Biosynthesis Defect with an Axonal Neuropathy and Metabolic Abnormality in Severe Cases. Am J Hum Genet. 2019;105(2):384-94.
11. Edvardson S, Murakami Y, Nguyen TTM, Shahrour M, St-Denis A, Shaag A, et al. Mutations in the phosphatidylinositol glycan C (PIGC) gene are associated with epilepsy and intellectual disability. J Med Genet. 2017;54(3):196-201.
12. Makrythanasis P, Kato M, Zaki MS, Saitsu H, Nakamura K, Santoni FA, et al. Pathogenic Variants in PIGG Cause Intellectual Disability with Seizures and Hypotonia. Am J Hum Genet. 2016;98(4):615-26.
13. Zhao JJ, Halvardson J, Knaus A, Georgii-Hemming P, Baeck P, Krawitz PM, et al. Reduced cell surface levels of GPI-linked markers in a new case with PIGG loss of function. Hum Mutat. 2017;38(10):1394-401.
14. Pagnamenta AT, Murakami Y, Anzilotti C, Titheradge H, Oates AJ, Morton J, et al. A homozygous variant disrupting the PIGH start-codon is associated with developmental delay, epilepsy, and microcephaly. Hum Mutat. 2018;39(6):822-6.
15. Nguyen TTM, Mahida S, Smith-Hicks C, Campeau PM. A PIGH mutation leading to GPI deficiency is associated with developmental delay and autism. Hum Mutat. 2018;39(6):827-9.
16. Nguyen TTM, Murakami Y, Mobilio S, Niceta M, Zampino G, Philippe C, et al. Bi-allelic Variants in the GPI Transamidase Subunit PIGK Cause a Neurodevelopmental Syndrome with Hypotonia, Cerebellar Atrophy, and Epilepsy. Am J Hum Genet. 2020;106(4):484-95.
17. Ng BG, Hackmann K, Jones MA, Eroshkin AM, He P, Wiliams R, et al. Mutations in the glycosylphosphatidylinositol gene PIGL cause CHIME syndrome. Am J Hum Genet. 2012;90(4):685-8.
18. Ceroni JR, Yamamoto GL, Honjo RS, Kim CA, Passos-Bueno MR, Bertola DR. Large deletion in PIGL: a common mutational mechanism in CHIME syndrome? Genet Mol Biol. 2018;41(1):85-91.
19. Fujiwara I, Murakami Y, Niihori T, Kanno J, Hakoda A, Sakamoto O, et al. Mutations in PIGL in a patient with Mabry syndrome. Am J Med Genet A. 2015;167A(4):777-85.
20. Pagnamenta AT, Murakami Y, Taylor JM, Anzilotti C, Howard MF, Miller V, et al. Analysis of exome data for 4293 trios suggests GPI-anchor biogenesis defects are a rare cause of developmental disorders. Eur J Hum Genet. 2017;25(6):669-79.
21. Mogami Y, Suzuki Y, Murakami Y, Ikeda T, Kimura S, Yanagihara K, et al. Early infancy-onset stimulation-induced myoclonic seizures in three siblings with inherited glycosylphosphatidylinositol (GPI) anchor deficiency. Epileptic Disord. 2018;20(1):42-50.
22. Altassan R, Fox S, Poulin C, Buhas D. Hyperphosphatasia with mental retardation syndrome, expanded phenotype of PIGL related disorders. Mol Genet Metab Rep. 2018;15:46-9.
23. Onoufriadis A, Simpson JK, McDonald C, Nguyen TTM, Campeau PM, Simpson MA, et al. Nonsyndromic erythrodermic ichthyosis resulting from a homozygous mutation in PIGL. Clin Exp Dermatol. 2020;45(3):391-4.
24. Almeida AM, Murakami Y, Layton DM, Hillmen P, Sellick GS, Maeda Y, et al. Hypomorphic promoter mutation in PIGM causes inherited glycosylphosphatidylinositol deficiency. Nat Med. 2006;12(7):846-51.
25. Pode-Shakked B, Heimer G, Vilboux T, Marek-Yagel D, Ben-Zeev B, Davids M, et al. Cerebral and portal vein thrombosis, macrocephaly and atypical absence seizures in Glycosylphosphatidyl inositol deficiency due to a PIGM promoter mutation. Mol Genet Metab. 2019;128(1-2):151-61.

26. Maydan G, Noyman I, Har-Zahav A, Neriah ZB, Pasmanik-Chor M, Yeheskel A, et al. Multiple congenital anomalies-hypotonia-seizures syndrome is caused by a mutation in PIGN. J Med Genet. 2011;48(6):383-9.
27. Ohba C, Okamoto N, Murakami Y, Suzuki Y, Tsurusaki Y, Nakashima M, et al. PIGN mutations cause congenital anomalies, developmental delay, hypotonia, epilepsy, and progressive cerebellar atrophy. Neurogenetics. 2014;15(2):85-92.
28. Khayat M, Tilghman JM, Chervinsky I, Zalman L, Chakravarti A, Shalev SA. A PIGN mutation responsible for multiple congenital anomalies-hypotonia-seizures syndrome 1 (MCAHS1) in an Israeli-Arab family. Am J Med Genet A. 2016;170A(1):176-82.
29. Nakagawa T, Taniguchi-Ikeda M, Murakami Y, Nakamura S, Motooka D, Emoto T, et al. A novel PIGN mutation and prenatal diagnosis of inherited glycosylphosphatidylinositol deficiency. Am J Med Genet A. 2016;170A(1):183-8.
30. Alessandri J-L, Gordon CT, Jacquemont M-L, Gruchy N, Ajeawung NF, Benoist G, et al. Recessive loss of function PIGN alleles, including an intragenic deletion with founder effect in La Réunion Island, in patients with Fryns syndrome. Eur J Hum Genet. 2018;26(3):340-9.
31. Krawitz PM, Murakami Y, Hecht J, Krüger U, Holder SE, Mortier GR, et al. Mutations in PIGO, a member of the GPI-anchor-synthesis pathway, cause hyperphosphatasia with mental retardation. Am J Hum Genet. 2012;91(1):146-51.
32. Nakamura K, Osaka H, Murakami Y, Anzai R, Nishiyama K, Kodera H, et al. PIGO mutations in intractable epilepsy and severe developmental delay with mild elevation of alkaline phosphatase levels. Epilepsia. 2014;55(2):e13-17.
33. Xue J, Li H, Zhang Y, Yang Z. Clinical and genetic analysis of two Chinese infants with Mabry syndrome. Brain Dev. 2016;38(9):807-18.
34. Tanigawa J, Mimatsu H, Mizuno S, Okamoto N, Fukushi D, Tominaga K, et al. Phenotype-genotype correlations of PIGO deficiency with variable phenotypes from infantile lethality to mild learning difficulties. Hum Mutat. 2017;38(7):805-15.
35. Morren M-A, Jaeken J, Visser G, Salles I, Van Geet C, NIHR BioResource, et al. PIGO deficiency: palmoplantar keratoderma and novel mutations. Orphanet J Rare Dis. 2017;12(1):101.
36. Zehavi Y, von Renesse A, Daniel-Spiegel E, Sapir Y, Zalman L, Chervinsky I, et al. A homozygous PIGO mutation associated with severe infantile epileptic encephalopathy and corpus callosum hypoplasia, but normal alkaline phosphatase levels. Metab Brain Dis. 2017;32(6):2131-7.
37. Holtz AM, Harrington AW, McNamara ER, Kielian A, Soul JS, Martinez-Ojeda M, et al. Expanding the phenotypic spectrum of Mabry Syndrome with novel PIGO gene variants associated with hyperphosphatasia, intractable epilepsy, and complex gastrointestinal and urogenital malformations. Eur J Med Genet. 2020;63(4):103802.
38. Johnstone DL, Nguyen T-T-M, Murakami Y, Kernohan KD, Tétreault M, Goldsmith C, et al. Compound heterozygous mutations in the gene PIGP are associated with early infantile epileptic encephalopathy. Hum Mol Genet. 2017;26(9):1706-15.
39. Krenn M, Knaus A, Westphal DS, Wortmann SB, Polster T, Woermann FG, et al. Biallelic mutations in PIGP cause developmental and epileptic encephalopathy. Ann Clin Transl Neurol. 2019;6(5):968-73.
40. Vetro A, Pisano T, Chiaro S, Procopio E, Guerra A, Parrini E, et al. Early infantile epileptic-dyskinetic encephalopathy due to biallelic PIGP mutations. Neurol Genet. 2020;6(1):e387.
41. Martin HC, Kim GE, Pagnamenta AT, Murakami Y, Carvill GL, Meyer E, et al. Clinical whole-genome sequencing in severe early-onset epilepsy reveals new genes and improves molecular diagnosis. Hum Mol Genet. 2014;23(12):3200-11.
42. Bellai-Dussault K, Nguyen TTM, Baratang NV, Jimenez-Cruz DA, Campeau PM. Clinical variability in inherited glycosylphosphatidylinositol deficiency disorders. Clin Genet. 2019;95(1):112-21.
43. Starr LJ, Spranger JW, Rao VK, Lutz R, Yetman AT. PIGQ glycosylphosphatidylinositol-anchored protein deficiency: Characterizing the phenotype. Am J Med Genet A. 2019;179(7):1270-5.
44. Nguyen TTM, Murakami Y, Wigby KM, Baratang NV, Rousseau J, St-Denis A, et al. Mutations in PIGS, Encoding a GPI Transamidase, Cause a Neurological Syndrome Ranging from Fetal Akinesia to Epileptic Encephalopathy. Am J Hum Genet. 2018;103(4):602-11.
45. Kvarnung M, Nilsson D, Lindstrand A, Korenke GC, Chiang SCC, Blennow E, et al. A novel intellectual disability syndrome caused by GPI anchor deficiency due to homozygous mutations in PIGT. J Med Genet. 2013;50(8):521-8.
46. Kohashi K, Ishiyama A, Yuasa S, Tanaka T, Miya K, Adachi Y, et al. Epileptic apnea in a patient with inherited glycosylphosphatidylinositol anchor deficiency and PIGT mutations. Brain Dev. 2018;40(1):53-7.
47. Yang L, Peng J, Yin X-M, Pang N, Chen C, Wu T-H, et al. Homozygous PIGT Mutation Lead to Multiple Congenital Anomalies-Hypotonia Seizures Syndrome 3. Front Genet. 2018;9:153.
48. Nakashima M, Kashii H, Murakami Y, Kato M, Tsurusaki Y, Miyake N, et al. Novel compound heterozygous PIGT mutations caused multiple congenital anomalies-hypotonia-seizures syndrome 3. Neurogenetics. 2014;15(3):193-200.
49. Lam C, Golas GA, Davids M, Huizing M, Kane MS, Krasnewich DM, et al. Expanding the clinical and molecular characteristics of PIGT-CDG, a disorder of glycosylphosphatidylinositol anchors. Mol Genet Metab. 2015;115(2-3):128-40.
50. Kawamoto M, Murakami Y, Kinoshita T, Kohara N. Recurrent aseptic meningitis with PIGT mutations: a novel pathogenesis of recurrent meningitis successfully treated by eculizumab. BMJ Case Rep. 2018;2018.
51. Bayat A, Knaus A, Juul AW, Dukic D, Gardella E, Charzewska A, et al. PIGT-CDG, a disorder of the glycosylphosphatidylinositol anchor: description of 13 novel patients and expansion of the clinical characteristics. Genet Med. 2019;21(10):2216-23.

52. Skauli N, Wallace S, Chiang SCC, Barøy T, Holmgren A, Stray-Pedersen A, et al. Novel PIGT Variant in Two Brothers: Expansion of the Multiple Congenital Anomalies-Hypotonia Seizures Syndrome 3 Phenotype. Genes (Basel). 2016;7(12).

53. Knaus A, Kortüm F, Kleefstra T, Stray-Pedersen A, Đukić D, Murakami Y, et al. Mutations in PIGU Impair the Function of the GPI Transamidase Complex, Causing Severe Intellectual Disability, Epilepsy, and Brain Anomalies. Am J Hum Genet. 2019;105(2):395-402.

54. Rabe P, Haverkamp F, Emons D, Rosskamp R, Zerres K, Passarge E. Syndrome of developmental retardation, facial and skeletal anomalies, and hyperphosphatasia in two sisters: nosology and genetics of the Coffin-Siris syndrome. Am J Med Genet. 1991;41(3):350-4.

55. Marcelis CL, Rieu P, Beemer F, Brunner HG. Severe mental retardation, epilepsy, anal anomalies, and distal phalangeal hypoplasia in siblings. Clin Dysmorphol. 2007;16(2):73-6.

56. Thompson MD, Roscioli T, Marcelis C, Nezarati MM, Stolte-Dijkstra I, Sharom FJ, et al. Phenotypic variability in hyperphosphatasia with seizures and neurologic deficit (Mabry syndrome). Am J Med Genet A. 2012;158A(3):553-8.

57. Krawitz PM, Schweiger MR, Rödelsperger C, Marcelis C, Kölsch U, Meisel C, et al. Identity-by-descent filtering of exome sequence data identifies PIGV mutations in hyperphosphatasia mental retardation syndrome. Nat Genet. 2010;42(10):827-9.

58. Horn D, Krawitz P, Mannhardt A, Korenke GC, Meinecke P. Hyperphosphatasia-mental retardation syndrome due to PIGV mutations: expanded clinical spectrum. Am J Med Genet A. 2011;155A(8):1917-22.

59. Horn D, Wieczorek D, Metcalfe K, Barić I, Paležac L, Cuk M, et al. Delineation of PIGV mutation spectrum and associated phenotypes in hyperphosphatasia with mental retardation syndrome. Eur J Hum Genet. 2014;22(6):762-7.

60. Reynolds KK, Juusola J, Rice GM, Giampietro PF. Prenatal presentation of Mabry syndrome with congenital diaphragmatic hernia and phenotypic overlap with Fryns syndrome. Am J Med Genet A. 2017;173(10):2776-81.

61. Foskett GK, Engleman E, Klotz J, Choi O, Tolentino L, Kochhar A, et al. Use of Flow Cytometry for Diagnosis of Epilepsy Associated With Homozygous PIGW Variants. Pediatr Neurol. 2018;85:67-70.

62. Chiyonobu T, Inoue N, Morimoto M, Kinoshita T, Murakami Y. Glycosylphosphatidylinositol (GPI) anchor deficiency caused by mutations in PIGW is associated with West syndrome and hyperphosphatasia with mental retardation syndrome. J Med Genet. 2014;51(3):203-7.

63. Hogrebe M, Murakami Y, Wild M, Ahlmann M, Biskup S, Hörtnagel K, et al. A novel mutation in PIGW causes glycosylphosphatidylinositol deficiency without hyperphosphatasia. Am J Med Genet A. 2016;170(12):3319-22.

64. Fu L, Liu Y, Chen Y, Yuan Y, Wei W. Mutations in the PIGW gene associated with hyperphosphatasia and mental retardation syndrome: a case report. BMC Pediatr. 27 2019;19(1):68.

65. Peron A, Iascone M, Salvatici E, Cavirani B, Marchetti D, Corno S, et al. PIGW-related glycosylphosphatidylinositol deficiency: Description of a new patient and review of the literature. Am J Med Genet A. 2020;

66. Ilkovski B, Pagnamenta AT, O'Grady GL, Kinoshita T, Howard MF, Lek M, et al. Mutations in PIGY: expanding the phenotype of inherited glycosylphosphatidylinositol deficiencies. Hum Mol Genet. 1 nov 2015;24(21):6146-59.

67. Murakami Y, Tawamie H, Maeda Y, Büttner C, Buchert R, Radwan F, et al. Null mutation in PGAP1 impairing Gpi-anchor maturation in patients with intellectual disability and encephalopathy. PLoS Genet. 2014;10(5):e1004320.

68. Bosch DGM, Boonstra FN, Kinoshita T, Jhangiani S, de Ligt J, Cremers FPM, et al. Cerebral visual impairment and intellectual disability caused by PGAP1 variants. Eur J Hum Genet. 2015;23(12):1689-93.

69. Kettwig M, Elpeleg O, Wegener E, Dreha-Kulaczewski S, Henneke M, Gärtner J, et al. Compound heterozygous variants in PGAP1 causing severe psychomotor retardation, brain atrophy, recurrent apneas and delayed myelination: a case report and literature review. BMC Neurol. 2016;16:74.

70. Hansen L, Tawamie H, Murakami Y, Mang Y, ur Rehman S, Buchert R, et al. Hypomorphic mutations in PGAP2, encoding a GPI-anchor-remodeling protein, cause autosomal-recessive intellectual disability. Am J Hum Genet. 2013;92(4):575-83.

71. Krawitz PM, Murakami Y, Rieß A, Hietala M, Krüger U, Zhu N, et al. PGAP2 mutations, affecting the GPI-anchor-synthesis pathway, cause hyperphosphatasia with mental retardation syndrome. Am J Hum Genet. 2013;92(4):584-9.

72. Jezela-Stanek A, Ciara E, Piekutowska-Abramczuk D, Trubicka J, Jurkiewicz E, Rokicki D, et al. Congenital disorder of glycosylphosphatidylinositol (GPI)-anchor biosynthesis--The phenotype of two patients with novel mutations in the PIGN and PGAP2 genes. Eur J Paediatr Neurol. 2016;20(3):462-73.

73. Naseer MI, Rasool M, Jan MM, Chaudhary AG, Pushparaj PN, Abuzenadah AM, et al. A novel mutation in PGAP2 gene causes developmental delay, intellectual disability, epilepsy and microcephaly in consanguineous Saudi family. J Neurol Sci. 2016;371:121-5.

74. Perez Y, Wormser O, Sadaka Y, Birk R, Narkis G, Birk OS. A Rare Variant in PGAP2 Causes Autosomal Recessive Hyperphosphatasia with Mental Retardation Syndrome, with a Mild Phenotype in Heterozygous Carriers. Biomed Res Int. 2017;2017:3470234.

75. Thompson MD, Knaus AA, Barshop BA, Caliebe A, Muhle H, Nguyen TTM, et al. A post glycosylphosphatidylinositol (GPI) attachment to proteins, type 2 (PGAP2) variant identified in Mabry syndrome index cases: Molecular genetics of the prototypical inherited GPI disorder. Eur J Med Genet. 2020;63(4):103822.

76. Howard MF, Murakami Y, Pagnamenta AT, Daumer-Haas C, Fischer B, Hecht J, et al. Mutations in PGAP3 impair GPI-anchor maturation, causing a subtype of hyperphosphatasia with mental retardation. Am J Hum Genet. 2014;94(2):278-87.

77. Knaus A, Awaya T, Helbig I, Afawi Z, Pendziwiat M, Abu-Rachma J, et al. Rare Noncoding Mutations Extend the Mutational Spectrum in the PGAP3 Subtype of Hyperphosphatasia with Mental Retardation Syndrome. Hum Mutat. 2016;37(8):737-44.

78. Abdel-Hamid MS, Issa MY, Otaify GA, Abdel-Ghafar SF, Elbendary HM, Zaki MS. PGAP3-related hyperphosphatasia with mental retardation syndrome: Report of 10 new patients and a homozygous founder mutation. Clin Genet. 2018;93(1):84-91.

79. Nampoothiri S, Hebbar M, Roy AG, Kochumon SP, Bielas S, Shukla A, et al. Hyperphosphatasia with Mental Retardation Syndrome Due to a Novel Mutation in PGAP3. J Pediatr Genet. 2017;6(3):191-3.

80. Sakaguchi T, Žigman T, Petković Ramadža D, Omerza L, Pušeljić S, Ereš Hrvaćanin Z, et al. A novel PGAP3 mutation in a Croatian boy with brachytelephalangy and a thin corpus callosum. Hum Genome Var. 2018;5:18005.

81. Akgün Doğan Ö, Demir GÜ, Kosukcu C, Taskiran EZ, Simsek-Kiper PÖ, Utine GE, et al. Hyperphosphatasia with mental retardation syndrome type 4 In two siblings-expanding the phenotypic and mutational spectrum. Eur J Med Genet. 2019;62(6):103535.

82. Balobaid A, Ben-Omran T, Ramzan K, Altassan R, Almureikhi M, Musa S, et al. Delineating the phenotypic spectrum of hyperphosphatasia with mental retardation syndrome 4 in 14 patients of Middle-Eastern origin. Am J Med Genet A. 2018;176(12):2850-7.

83. Abi Farraj L, Khatoun WD, Abou Chebel N, Wakim V, Dawali K, Ghassibe-Sabbagh M. Clinical, genetic, and molecular characterization of hyperphosphatasia with mental retardation: a case report and literature review. Diagn Pathol. 2019;14(1):123.

84. Nguyen TTM, Murakami Y, Sheridan E, Ehresmann S, Rousseau J, St-Denis A, et al. Mutations in GPAA1, Encoding a GPI Transamidase Complex Protein, Cause Developmental Delay, Epilepsy, Cerebellar Atrophy, and Osteopenia. Am J Hum Genet. 2017;101(5):856-65.