

Figure S1: Venn diagram representing overlaps of genes associated with the indicated phenotypic abnormalities in the Synthesis group.



Figure S2: Venn diagram representing genes found in at diseases that overlap by at least 14 phenotypic abnormalities. Two genes, FGFR2 and BRAF, were associated with Mendelian diseases that are characterized by all 15 phenotypic features. KRAS was associated with the 13 phenotypes as well as Macrotia. The 8 genes shown were listed with the 13 phenotypic features and Sleep disturbance. The 13 phenotypes were: Abnormal bone density, Abnormal bone ossification, Abnormal bone structure, Abnormality of nose, Behavioral abnormality, Cleft palate, Hard palate, Intellectual disability, Neurodevelopmental delay, Neurodevelopmental abnormality, Oral cleft, Osteopenia, Reduced bone density.

Table **S1**: Case reports included in the present work. The gene, patient id, and a list of the HPO terms that we curated for each patient are shown. S: Synthesis group. T+R: Transamidase+Remodeling group.

Gene	Patient	HPOs				Group	PMID
	ID					-	
DPM1	PY	HP:0012448,	HP:0004322,	HP:0001643,	HP:0100704,	S	10642597
		HP:0002015,	HP:0002104,	HP:0001250,	HP:0000822,		
		HP:0001272,	HP:0000486,	HP:0003186,	HP:0005484,		
		HP:0003236,	HP:0007333	,	,		
DPM1	CH	HP:0012448,	HP:0006801,	HP:0000174,	HP:0002353,	\mathbf{S}	10642597
		HP:0010844,	HP:0002374,	HP:0100704,	HP:0001250,		
		HP:0011398,	HP:0001263,	HP:0000961,	HP:0005484,		
		HP:0001028,	HP:0005949,	HP:0000494,	HP:0003259,		
		HP:0001009,	HP:0000271	,	,		
DPM1	Patient	HP:0001270,	HP:0001250,	HP:0011398,	HP:0002457,	\mathbf{S}	23856421
		HP:0005484,	HP:0012385, HF	2:0009276, HP:0	040081		
DPM2	Patient 3	HP:0001999,	HP:0002058,	HP:0002650,	HP:0002421,	\mathbf{S}	23109149
		HP:0002197,	HP:0011968,	HP:0011947,	HP:0040081,		
		HP:0002803,	HP:0030951,	HP:0001250,	HP:0011398,		
		HP:0001263,	HP:0003198,	HP:0001321,	HP:0001320,		
		HP:0001344,	HP:0005484	,	,		
DPM2	Patient 2	HP:0001999,	HP:0002058,	HP:0002650,	HP:0002421,	\mathbf{S}	23109149
		HP:0002197,	HP:0011968,	HP:0011947,	HP:0040081,		
		HP:0002803,	HP:0030951,	HP:0000689,	HP:0001250,		
		HP:0000347,	HP:0011398,	HP:0001263,	HP:0000486,		
		HP:0003198,	HP:0001321,	HP:0001320,	HP:0001344,		
		HP:0005484					
DPM2	Patient 1	HP:0002194,	HP:0025404,	HP:0002421,	HP:0012157,	S	23109149
		HP:0002205,	HP:0011947,	HP:0000617,	HP:0002910,		
		HP:0000219,	HP:0002705,	HP:0006380,	HP:0001250,		
		HP:0000347,	HP:0003196,	HP:0011398,	HP:0011344,		
		HP:0001561,	HP:0005484,	HP:0100360,	HP:0000294,		
		HP:0008936,	HP:0002240,	HP:0002002,	HP:0040246,		
		HP:0002553,	HP:0002098,	HP:0030903,	HP:0030235,		
		HP:0002987,	HP:0002878,	HP:0000648,	HP:0010460,		
		HP:0100543,	HP:0000601, HF	P:0000271, HP:0	002090		
DPM3	Patient	HP:0004302,	HP:0003805,	HP:0004322,	HP:0003707,	\mathbf{S}	19576565
		HP:0025502,	HP:0001644,	HP:0001763,	HP:0003160,		
		HP:0002401,	HP:0002910,	HP:0002515,	HP:0003487,		
		HP:0005109,	HP:0003560,	HP:0003198,	HP:0003701,		
		HP:0009053,	HP:0001928,	HP:0003259,	HP:0003555,		
		HP:0001315					
DPM3	Patient	HP:0004302,	HP:0003805,	HP:0004322,	HP:0003707,	\mathbf{S}	19576565
		HP:0025502,	HP:0001644,	HP:0001763,	HP:0003160,		
		HP:0002401,	HP:0002910,	HP:0002515,	HP:0003487,		
		HP:0005109,	HP:0003560,	HP:0003198,	HP:0003701,		
		HP:0009053,	HP:0001928,	HP:0003259,	HP:0003555,		
		HP:0001315					

Gene	Patient ID	HPOs				Group	\overline{PMII}
MPDU1	Patient S	HP:0040189,	HP:0001250,	HP:0008064,	HP:0003510,	S	1173356
		HP:0011344,	HP:0001541,	HP:0001276,	HP:0011968,		
		HP:0001510,	HP:0012050,	HP:0040189,	HP:0040196,		
		HP:0001698,	HP:0001873,	HP:0011968,	HP:0002119,		
		HP:0002521,	HP:0000803,	HP:0000648,	HP:0012050,		
		HP:0001250,	HP:0003510,	HP:0008064,	HP:0011344,		
		HP:0001541,	HP:0001276,	HP:0001510,	HP:0012704,		
		HP:0040189,	HP:0001319,	HP:0011968,	HP:0012050,		
		HP:0001250,	HP:0003510,	HP:0008064,	HP:0011344,		
		HP:0001541,	HP:0000233,	HP:0001276,	HP:0000242,		
		HP:0001510,	HP:0000260				
PIGA	III-7	HP:0012469,	HP:0001263, HI	P:0002521		\mathbf{S}	2435751
	Family B						
PIGA	III-13	HP:0001250,	HP:0001263, HI	P:0002521, HP:0	0011947	\mathbf{S}	2435751
	Family B						
PIGA	III-11	HP:0002133,	HP:0002069,	HP:0001263,	HP:0002521,	\mathbf{S}	2435751
	Family B	HP:0000969					
PIGA	III-8	HP:0012469,	HP:0001272,	HP:0001263,	HP:0002521,	\mathbf{S}	2435751
	Family B	HP:0002500,	HP:0002090				
PIGA IV-2	IV-2	HP:0001270,	HP:0008936,	HP:0001250,	HP:0001263,	\mathbf{S}	2435751
	Family B	HP:0002521,	HP:0001336				
PIGA IV-4	HP:0002079,	HP:0002123,	HP:0001623,	HP:0001643,	\mathbf{S}	2230553	
	HP:0002878,	HP:0002139,	HP:0003155,	HP:0002705,			
		HP:0001371,	HP:0001252,	HP:0001561,	HP:0001321,		
		HP:0025116,	HP:0011330, H	P:0001414, HP:0	0001348	-	
PIGA	IV-2	HP:0002079,	HP:0006956,	HP:0005280,	HP:0001623,	\mathbf{S}	2230553
		HP:0000076,	HP:0001631,	HP:0001792,	HP:0002104,		
		HP:0002714,	HP:0006380,	HP:0003273,	HP:0000212,		
		HP:0001321,	HP:0001331,	HP:0009381,	HP:0001169,		
		HP:0000272,	HP:0000470,	HP:0000160,	HP:0002123,		
		HP:0002987,	HP:0000239,	HP:0000269,	HP:0000201,		
		HP:0000463,	HP:0000396,	HP:0000582,	HP:0001348,		
		HP:0002090,	HP:0000081			-	
PIGA	IV-4	HP:0002123,	HP:0002079,	HP:0001250,	HP:0001371,	\mathbf{S}	2470601
		HP:0010851,	HP:0011398,	HP:0000076,	HP:0001347,		
		HP:0012430,	HP:0000271,	HP:0003155,	HP:0002123,		
		HP:0002079,	HP:0001371,	HP:0010851,	HP:0011398,		
		HP:0001561,	HP:0001367,	HP:0001347,	HP:0012430,		
DIGA	2	HP:0000271			HD 0011000	a	0.1-0.001
PIGA	5	HP:0002187,	HP:0003155,	HP:0002079,	HP:0011398,	S	2470601
		HP:0002521,	HP:0001347, HI	P:0002445, HP:0	0000271	G	0.450.001
PIGA	4	HP:0003155,	HP:0002079,	HP:0001250,	HP:0001371,	S	2470601
		HP:0002342,	HP:0001367,	HP:0002510,	HP:0002521,		
	2	HP:0003429,	HP:0000271, H	P:0000717		C	0.450.001
PIGA	3	HP:0002187,	HP:0002197, HI	P:0010818, HP:0	0000717	S	2470601
PIGA	2	HP:0002187,	HP:0003155,	HP:0002123,	HP:0002079,	5	2470601
		HP:0001250,	HP:0001367,	HP:0002521,	HP:0002510,		
	1	HP:0001336,	пР:0012430, HI	r:0000271	IID.0001971	C	0.470001
FIGA	T	HP:0002187,	HP:0002079, $IID 0000076$	HP:0010851,	HP:0001371,	5	2470601
		HP:0011398,	HP:000076,	HP:0001561,	HP:0005692,		
	IV 9	HP:0012430,	пР:0002445, HI	r:0000271, HP:0	JUIU818	C	0470001
FIGA	1V-2	HP:0002123,	HP:0002079,	HP:0001371,	HP:0001250,	2	2470601
		HP:0010851,	HP:0011398,	HP:0000076,	нP:0001347,		
		HP:0012430.	6P:000271				

Table S1 – Continued from previous page

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Table S	51 -	Continued	trom	previous	paae
				P	1

Gene	Patient	HPOs		v <u>1</u>		Group	PMID
	ID					1	
PIGA	IV-3	HP:0001939,	HP:0003121,	HP:0001433,	HP:0003487,	S	24259288
		HP:0003700,	HP:0001257,	HP:0001397,	HP:0000365,		
		HP:0002078,	HP:0000496,	HP:0002240,	HP:0000684,		
		HP:0001298.	HP:0001251.	HP:0000248.	HP:0001332.		
		HP:0003281.	HP:0100704.	HP:0001250.	HP:0011398.		
		HP·0040130	111 10100101,		,		
PIGA	III-10	HP·0040134	HP·0001712	HP·0001744	HP·0002174	S	24259288
11011		HP.0012103	HP·0001873	HP.0001760	HP.0007190	~	_1_00_00
		HP:0200034	HP-0009909	HP:0002529	HP.0001250		
		HP·0001371	HP.0011952	HP:0000684	HP.0001200, HP.0001541		
		HP:0001298	HP:0005484	HP:0001399	HP:0000496		
		HP:0001939	HP:0001357	HP:0002240	HP:0002133		
		HP:0006349	HP:0004322	HP:0002650	HP:0100704		
		HP:0002753	HP:0002119	HP:0002590,	HP:0000029		
		HP-0003202	HP:0000687	HP:00012557	HP:0000601		
		HP:0003265	HD:0000007,	2.0001413 HP.0	111.0000000000000000000000000000000000		
PICA	111.0	HP-0002133	HD-0008036	HP-0100704	HP-0002171	S	24250288
IIGA	111-3	HP.0002133,	HP:0030003	HP.0001182	HP.0011051	5	24209200
		HP.0002340, HP.0001250	HP.0000365	HP: 0001182 ,	HP.00011951,		
		HD.0001230,	HD:0001336	HD.0001044,	HD.0001248		
		UD.0001433,	111.0001330,	111 .0009085,	111.0001348,		
DICA	11.9	ПГ.0000280 ПР.0002192	HD-0001520	UD:0019726	UD.0019295	C	95995597
FIGA	11.2	11F.0002123, 11D.0001271	11F.0001520, 11D.0002226	11F.0012730, 11D.0006101	11F.0012363, 11D.0005484	a	20000021
		11F.0001371,	IIF:0002230, IID:0019745	11F.0000191, 11D.0000294	11F.00003464, 11D.0019460		
		$\Pi P:0002003,$ $\Pi D:0000627$	$\Pi P:0012743,$ $\Pi D:0008026$	$\Pi P:0000524,$ $\Pi D:0004672$	$\Pi P:0012409,$ $\Pi D:0005080$		
		$\Pi P:0000037,$	ПР:0008950, ПР:0009705	$\Pi P:0004075,$ $\Pi D:0000216$	$\Pi P:0000989,$ $\Pi D:0009967$		
		$\Pi P:0000248,$ $\Pi D:0002521$	$\Pi P:0002703,$ $\Pi D:0001950$	$\Pi P:0000310,$ $\Pi D:0000474$	$\Pi P:0002207,$ $\Pi D:0001257$		
		$\Pi P:0002521,$	$\Pi P:0001230,$ $\Pi D:0000211$	$\Pi P:0000474,$ $\Pi D:0000227$	$\Pi P:0001557,$ $\Pi D.0100729$		
		HP:0003409, HD:0002056	ПР:0000511, ПР:0001489	ПР:0000337, ПР:0002825	HP:0100338, HP:0000545		
		$\Pi P:0002030,$ $\Pi D:0004208$	$\Pi P:0001466,$ $\Pi D:0002050$	ПР:0002855, ПР:0000265	$\Pi P:0000343,$ $\Pi D:0000126$		
		$\Pi P:0004508,$ $\Pi D:0004446$	HP:0002039,	$\Pi P:0000505,$ $\Pi D:0009104$	$\Pi P:0000120,$ $\Pi D:0002240$		
		$\Pi P:0004440,$ $\Pi D:0001667$	$\Pi P:0000994,$	$\Pi P:0002104,$ $\Pi D:0002155$	$\Pi P:0002240,$ $\Pi D:0007270$		
		$\Pi P:0001007,$ $\Pi D:0020515$	$\Pi P:0000545,$ $\Pi D:0000556$	$\Pi P:0003133,$ $\Pi D:0001102$	$\Pi P:0007570,$ $\Pi D:0021145$		
		HP:0030515, HP:0031145	HP:0000550,	HP:0001103,	HP:0031145,		
PICA	TTT 1	HP:0000800	HD:0002070	HD:0002271	HD:0002376	S	24250184
IIGA	111-1	HP:0001631	HP:0001605	HP:0001671	HP:0002070,	5	24203104
		HD.0001031,	HD.0001055,	HD.0001071,	HD.0002009,		
		HP.0002104,	HP:001250,	HP.0012420	HP.0001301,		
		HP:0008936	HP:0002764,	$HP \cdot 0002425,$	HP:0002373,		
		HP-0002119	HP:0002200,	$HP \cdot 0002155$	HP:0001263		
		HP:0200134	HD:0000270,	2.000000000000000000000000000000000000	111.0001203,		
PICA	Oldor	HP-0002133	HP-0005280	HP-0002123	HP:0002	S	20502866
IIGA	twin	HP.0002155,	HP:0002491	HP:0025404	HP:0002376	5	29502800
	6 W 111	HD.0002009,	HD.0002421,	HD.00023404, HD.0000316	HP.0002370,		
		HD:0012001,	пп.0000017, чр.0001252 нг	2.0000463	111.0001250,		
DICA	Vouncor	ПП.0011108, 1 П.0002122	ир.0001252, III	UD:0002170	UD.0002060	C	20502266
FIGA	turin	IIF.0002133, IID.0002431	HP:0005280,	ПГ:0002179, ПР:0002276	ПР:0002009, ПР:0012001	a	29302800
	UWIII	HP.0002421, HP.0000817	HP:0000316	HP:0002370,	HP.00012001, HP.0001252		
		HP.0000817,	111.0000310,	111.0001250,	111.0001252,		
DICC	Family	HP.0001270	UD.0002060	UD:0001950	UD.0001962	C	27604521
гюU	гашцуА П 4	HD.00100270,	11F:0002009, HD:00007F0 III	11F:0001200, 0.0021401	111:0001203,	S	21094321
DICC	11-4 Fomile-A	HD.0001970	HD.0002060	.0031491 HD.0001950	HD.0001969	C	97604591
гюU	ганнуА 11 э	11F:0001270, 11D:0010967	11F:0002009, UD:0000780 III	11F:0001200, 0.0021401 11D:0	11F:0001203,	S	21094321
DICC	II-Z	пг:0010804, I	HE:UUUU/80, HE	∴0051491, HP:(2.0001240	100702	C	97604591
FIGU	гашцув 11-1	hF:0001250, I	117:0001203, HI	0001249		ъ	27094321
	11-1						

Table S1 – Continued from previous page

Gene	Patient	HPOs				Group	PMID
- DTG G	ID		TTD 000 (000	TTD 0004000		~	
PIGG	JP01	HP:0001250,	HP:0004396,	HP:0001263,	HP:0000750,	S	26996948
		HP:0001252,	HP:0001321,	HP:0001510,	HP:0002059,		
DIGG	DUGA	HP:0000717				a	0.000.00.40
PIGG	PK01	HP:0001263,	HP:0001250,	HP:0006829,	HP:0007370,	S	26996948
		HP:0011343,	HP:0001344,	HP:0000729,	HP:0008855,		
DIGG	DIZOO	HP:0030047				a	00000010
PIGG	PK02	HP:0001263,	HP:0001250,	HP:0006829,	HP:0007370,	S	26996948
		HP:0011343,	HP:0001344,	HP:0000729,	HP:0008855,		
DIGG	EC01	HP:0030047	UD 0001909	UD 0011900	IID 0011949	a	0000040
PIGG	EG01	HP:0001250,	HP:0001382,	HP:0011398,	HP:0011343,	S	26996948
		HP:0001263,	HP:0006829,	HP:0007370,	HP:0000729,		
DIGG	ECOO	HP:0001344,	HP:0008855, HF	2:0030047	UD 0001069	C	0000040
PIGG	EG02	HP:0001250,	HP:0001382,	HP:0011343,	HP:0001263,	5	26996948
		HP:0006829,	HP:0007370,	HP:0000729,	HP:0001344,		
DICII	Duchaud	HP:0008855, UD:0009272	HP:0030047	IID.0009911	UD-0010949	C	90609516
PIGH	Proband	HP:0002373, UD:0000717	HP:0045045,	HP:0002311,	HP:0012343, HD:0011242	5	29003510
		HP:0000717,	HP:0001290,	HP:0000218,	HP:0011343,		
DICI	Detient	HP:0000750,	HP:0010529, HF	10001007	UD.0009644	C	20472027
PIGL	Patient	$\Pi P:0100800,$ $\Pi D:0009126$	$\Pi P:0002098,$ $\Pi D:0000216$	$\Pi P:0000120,$ $\Pi D:0002155$	$\Pi P:0002044,$ $\Pi D:0001250$	S	29473937
	10	$\Pi P:0002150,$ $\Pi D:0001962$	$\Pi P:0000510,$ $\Pi D:0001156$	$\Pi P:0003133,$ $\Pi D:0000079$	$\Pi P:0001230,$ $\Pi D:0008070$		
		$\Pi P:0001205,$ $\Pi D:0000206$	$\Pi P:0001130,$ $\Pi D:0007502$ III	$\Pi P:0000972,$	$\Pi P:0008070,$		
DICI	a in l	HP:0000590,	пг:0007505, пг ир.0005280	ПР:000480, ПР:0	JU01249 JID.0009491	C	25706256
PIGL	giri	HP:0000950, HD:0009750	$\Pi P:0005280,$ $\Pi D:0010546$	$\Pi P:0002194,$ $\Pi D:0004049$	$\Pi P:0002421,$ $\Pi P:0001702$	S	29700590
		HP.001102750,	HP.0010340, HP.0000627	IIF:0004042, UD:0000218	ПР:0001792, ПР:0001250		
		ПГ:0011927, ПО:0011208	HP.0000684	HP.0000218,	HP:0001250,		
		HP:0011398,	HP:0000280	HD:0000158	HP:0000303,		
		HP:0000316	HP:000280,	HP:00011800	HP.0002789, HP.0001320		
		HP:0000310,	HP:0003155,	HP: 0011800 ,	HP.0001320, HP.0000203		
		HP:0006056	HP.0005280	HP:0001344, HP:0002104	HP:000295,		
		HP:0002750	HP:0010546	HP: 0002194 ,	HP.0002421, HP.0001702		
		HP.001102750,	HP.0000637	HP:0004042,	HP.0001792,		
		HP.0011327,	HP.0000684	HP:0000218,	HP:0001250,		
		HP:0010804	HP:0000280	$HP \cdot 0000480,$	HP:0002780		
		HP:0000316	HP:0003155	$HP \cdot 0011800$	HP.0002789, HP.0001320		
		HP:0000364	HP-0000463 HF	2.0001344 HP·0	0001020,		
PIGL	3988	HP:0400005	HP·0010783	HP-0005280	HP-0004209	S	22444671
1101	0000	HP:0001682	HP:0000126	HP:00009200,	HP:0002136	D	22111011
		HP.0011927	HP:0002007	HP:0000405	HP:0000589		
		HP:0001250	HP:0000369	HP:0000358	HP:0008064		
		HP:0000343	HP·0000271 HF	P·0001019			
PIGL	0680-2-3	HP:0000175.	HP:0002286.	HP:0002213.	HP:0012471.	S	22444671
1101	0000 2 0	HP:0000316	HP:0000589	HP:0001250	HP.0000248	N	22111011
		HP:0008064.	HP:0011069.	HP:0000687.	HP:0000972.		
		HP:0000431.	HP:0001288.	HP:0008591.	HP:0008070.		
		HP:0000396.	HP:0000286.	HP:0100040.	HP:0001249.		
		HP:0001638	,	,	,		
PIGL	0680-2-2	HP:0000175,	HP:0002286,	HP:0002213,	HP:0002059,	\mathbf{S}	22444671
		HP:0012471.	HP:0000316.	HP:0000457.	HP:0000589,		
		HP:0001250,	HP:0000248,	HP:0008064,	HP:0011069,		
		HP:0000687,	HP:0000972,	HP:0000431,	HP:0001288,		
		HP:0008591,	HP:0008070,	HP:0000396,	HP:0000286,		
		HP:0100040,	HP:0001249, HF	2:0001638	,		
			~				

Gene	Patient ID	HPOs				Group	PMID
PIGL	33300	HP:0010047,	HP:0000193,	HP:0005280,	HP:0010049,	S	22444671
		HP:0100759,	HP:0025474,	HP:0002213,	HP:0002059,		
		HP:0000958,	HP:0100542,	HP:0100760,	HP:0001270,		
		HP:0000589,	HP:0008064,	HP:0000894,	HP:0000377,		
		HP:0008070,	HP:0009931,	HP:0000286,	HP:0000252,		
		HP:0001169,	HP:0001999,	HP:0001769,	HP:0001945,		
		HP:0010783,	HP:0004279,	HP:0001943,	HP:0010038,		
		HP:0012211,	HP:0000989,	HP:0002901,	HP:0012471,		
		HP:0030148,	HP:0005469,	HP:0000405,	HP:0000316,		
		HP:0000455,	HP:0007808,	HP:0001156,	HP:0000322,		
		HP:0000972, HP:0001805	HP:0000463,	HP:0001598,	HP:0100040,		
PIGL	0682 - 2 - 1	HP:0002286,	HP:0002213,	HP:0002059,	HP:0012471,	\mathbf{S}	22444671
		HP:0000405,	HP:0000316,	HP:0000589,	HP:0001250,		
		HP:0000248,	HP:0008064,	HP:0011069,	HP:0000687,		
		HP:0000972,	HP:0000431,	HP:0001288,	HP:0008070,		
		HP:0000396, HP:0001638	HP:0000286,	HP:0100040,	HP:0001249,		
PIGL	277013	HP:0002069,	HP:0002194,	HP:0000154,	HP:0030084,	\mathbf{S}	28327575
		HP:0006347,	HP:0010055,	HP:0000107,	HP:0000958,		
		HP:0003155,	HP:0000405,	HP:0000965,	HP:0000767,		
		HP:0001263,	HP:0000687,	HP:0000365,	HP:0009381,		
		HP:0003764					
PIGM	Family 2- 2B	HP:0002121,	HP:0030242, HI	P:0030243		S	16767100
PIGM	Family 2- 2C	HP:0002121,	HP:0030242, HI	P:0030243		\mathbf{S}	16767100
PIGM	Family 1- 1B	HP:0002121,	HP:0030242, HI	P:0030243		\mathbf{S}	16767100
PIGN	Patient	HP:0001626,	HP:0002089,	HP:0001831,	HP:0030030,	\mathbf{S}	24852103
		HP:0000054,	HP:0000175,	HP:0000110,	HP:0012165,		
		HP:0002623,	HP:0002566,	HP:0000028,	HP:0001060,		
		HP:0025193,	HP:0003244,	HP:0000776,	HP:0000316,		
		HP:0000445,	HP:0000377, HI	P:0000463, HP:0	0000271		
PIGN	COLL- 2.3	HP:0000174,	HP:0000110, HI	P:0000803, HP:0	0012718	S	27038415
PIGN	PIGN;V-	HP:0001615,	HP:0002100,	HP:0000072,	HP:0001631,	\mathbf{S}	21493957
	2	HP:0000034,	HP:0000639,	HP:0001290,	HP:0000219,		
		HP:0000646,	HP:0002705,	HP:0000400,	HP:0000278,		
		HP:0000486,	HP:0000341,	HP:0001347,	HP:0001337,		
		HP:0011271,	HP:0001945,	HP:0000194,	HP:0002020,		
		HP:0002286,	HP:0000126,	HP:0000319,	HP:0002305,		
		HP:0010880,	HP:0000316,	HP:0001182,	HP:0000269,		
		HP:0001263,	HP:0002090	,	,		
PIGN	V-1	HP:0004467,	HP:0000194,	HP:0001655,	HP:0000034,	\mathbf{S}	21493957
		HP:0004969,	HP:0000639,	HP:0000646,	HP:0002705,		
		HP:0000347,	HP:0000269,	HP:0001156,	HP:0009748,		
		HP:0000774,	HP:0000341, HI	P:0010804, HP:0	0000280		
PIGN	NSGC- 74	HP:0000174,	HP:0000110, HI	P:0000803, HP:0	0012718	S	27038415
PIGN	Patient 1	HP:0002384	HP:0012448	HP:0000076	HP:0010544	S	24253414
	1 6010110 1	HP:0000010	HP:0000639	HP:0012381	HP:0002705	2	21200114
		HP:0001272	HP:0000347	HP:0011398	HP:0000341		
		HP:0003700	HP:0000286	HP:0010804	HP:0003324		
		HP:0001337	HP:0000194	HP:0004396	HP:0002119		
		UD.0000260	IID.0001969 III	2.0001944 IID.(010506		

Table S1 - Continued from previous page

Gene	Patient ID	HPOs				Group	PMII
PIGN	Patient2	HP:0002384,	HP:0012448,	HP:0000076,	HP:0010544,	S	2425341
		HP:0000010,	HP:0000639,	HP:0012381,	HP:0002705,		
		HP:0001272,	HP:0000347,	HP:0011398,	HP:0000341,		
		HP:0003700,	HP:0000286,	HP:0010804,	HP:0003324,		
		HP:0001337,	HP:0000194,	HP:0004396,	HP:0002119,		
		HP:0000269,	HP:0001263, HI	P:0001344, HP:0	0010506		
PIGN	male	HP:0002384,	HP:0012703,	HP:0000174,	HP:0002194,	\mathbf{S}	2909660
		HP:0002421,	HP:0002353,	HP:0002457,	HP:0002533,		
		HP:0004305,	HP:0003196,	HP:0001263,	HP:0100370,		
		HP:0100702,	HP:0001344, HI	P:0100371			
PIGN	NSGC- 7.3	HP:0000174,	HP:0000110, HI	P:0000803, HP:0	0012718	S	2703841
PIGN	Patient 2	HP:0006855,	HP:0006956,	HP:0100660,	HP:0002123,	\mathbf{S}	2687944
		HP:0001272,	HP:0025439,	HP:0001250,	HP:0010544,		
		HP:0000010,	HP:0001336, HI	P:0001337, HP:0	0001290		
PIGN	259633	HP:0100543,	HP:0001250,	HP:0012444,	HP:0002194,	\mathbf{S}	2832757
		HP:0007308,	HP:0002205, HI	P:0001344	1		
PIGO	II-1 fam-	HP:0003155,	HP:0001252,	HP:0001263,	HP:0010804,	S	2268308
	ilv A	HP:0002025,	HP:0000316,	HP:0000750,	HP:0001270,		
0	HP:0004322,	HP:0000455,	HP:0000431,	HP:0006118,			
	HP:0000076,	HP:0000637, HI	P:0002251	1			
PIGO II-2 fam-	HP:0003155,	HP:0001252,	HP:0001263,	HP:0010804,	\mathbf{S}	2268308	
	ilv A	HP:0002025,	HP:0000316,	HP:0004322,	HP:0000750,		
	HP:0001270,	HP:0000455,	HP:0000431,	HP:0006118,			
		HP:0000076.	HP:0000637	,			
PIGO	II-1 fam-	HP:0001250,	HP:0002265,	HP:0001263,	HP:0011316,	S	2268308
	ilv B	HP:0004969,	HP:0000324,	HP:0010804,	HP:0000316,		
	5	HP:0002540,	HP:0001270,	HP:0000637,	HP:0000494,		
		HP:0000508,	HP:0001792,	HP:0003155,	HP:0004209,		
		HP:0000455,	HP:0009642,	HP:0000431,	HP:0001631,		
		HP:0006118,	HP:0001800, HI	P:0000252, HP:0)009909		
PIGO	Boy	HP:0009882,	HP:0002251,	HP:0001636,	HP:0002134,	S	2404913
	- 0	HP:0008527.	HP:0012501.	HP:0002871.	HP:0002352.		
		HP:0002104.	HP:0003155.	HP:0001272.	HP:0011398.		
		HP:0000365.	HP:0000297.	HP:0001344.	HP:0009381.		
		HP:0003429,	HP:0031165, HI	P:0000271	1		
PIGO	Individual	HP:0002384.	HP:0002069.	HP:0002063.	HP:0010864.	S	2441774
	II-1	HP:0002510.	HP:0002059,	HP:0003155.	HP:0002705.		
		HP:0001250.	HP:0001263.	HP:0200134,	HP:0001252,		
		HP:0002072,	HP:0010804, HI	P:0100275	,		
PIGO	Individual II-2	HP:0002069,	HP:0001263			S	2441774
PIGO	263039	HP:0002251,	HP:0002013,	HP:0001810,	HP:0002164,	\mathbf{S}	2832757
		HP:0000407,	HP:0000219,	HP:0001270,	HP:0003155,		
		HP:0001263,	HP:0001156,	HP:0000364,	HP:0001344,		
		HP:0000252,	HP:0001817, HI	P:0001804	,		
PIGO	Patient 1	HP:0002384,	HP:0002100,	HP:0008936,	HP:0002079,	\mathbf{S}	2890081
		HP:0002120,	HP:0011968,	HP:0000817,	HP:0002307,		
		HP:0004305,	HP:0000758,	HP:0008763,	HP:0002835,		
		HP:0000711,	HP:0001263.	HP:0003560.	HP:0001298,		
		HP:0002093.	HP:0002090	;	,		
PIGO	Patient 2	HP:0012448.	HP:0002100.	HP:0008936.	HP:0002079.	\mathbf{S}	2890081
		HP:0012736.	HP:0001250.	HP:0001263,	HP:0002353.		
		HP-0000961	HP-0002120 HI	P-0002307 HP-0	010818		

Table S1 – Continued from previous page

$\mathbf{J}_{\mathbf{a}}$	Table $\mathbf{S1}$	- Continued	from	previous	page
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ID PIGP Male HP:0002187, HP:0002079, HP:0002353, HP:0010546, S 2835 purchand HD:0002457, HD:0002695, HD:0002353, HD:0002350, S 2835	84793
PIGP Male HP:0002187, HP:0002079, HP:0002353, HP:0010546, S 2833	34793
makend $IID.0009447$ $IID.0006004$ $IID.0006094$ $IID.0006094$	
$ proband \qquad HF: 0002457, \qquad HF: 0000895, \qquad HF: 0002835, \qquad HF: 00005380, $	
$\mathrm{HP:}0003037, \mathrm{HP:}0001250, \mathrm{HP:}0011398, \mathrm{HP:}0000961,$	
$\mathrm{HP:}0001344, \mathrm{HP:}0001357, \mathrm{HP:}0002094, \mathrm{HP:}0001618,$	
HP:0200049	
PIGP Female HP:0002100, HP:0010553, HP:0001250, HP:0002353, S 2833	34793
proband HP:0100704, HP:0000817, HP:0001347, HP:0002521,	
HP:0002169, HP:0002835, HP:0000549	
PIGQ Patient HP:0010851, HP:0001371, HP:0012443, HP:0001250 S 2446	53883
PIGV Patient 5 HP:0000750, HP:0011344, HP:0001792, HP:0009381, S 2412	29430
$\mathrm{HP:}0003155, \mathrm{HP:}0010804, \mathrm{HP:}0000283, \mathrm{HP:}0009882,$	
HP:0001831, HP:0000271, HP:0001336	
PIGV Patient 8 HP:0002251, HP:0000072, HP:0002025, HP:0006118, S 2412	29430
$\mathrm{HP:}0000076, \mathrm{HP:}0000021, \mathrm{HP:}0001792, \mathrm{HP:}0000126,$	
$\mathrm{HP:}0010055, \mathrm{HP:}0003155, \mathrm{HP:}0001250, \mathrm{HP:}0011344,$	
$\mathrm{HP:}0000750, \mathrm{HP:}0000431, \mathrm{HP:}0001344, \mathrm{HP:}0009381,$	
HP:0010804, HP:0000271	
PIGV Patient 9 HP:0001804, HP:0000455, HP:0001821, HP:0003155, S 2412	29430
$\mathrm{HP:}0000316, \mathrm{HP:}0000126, \mathrm{HP:}0200007, \mathrm{HP:}0010804,$	
HP:0000431, HP:0000175, HP:0001629, HP:0000072	
PIGV Patient 1 HP:0040255, HP:0002023, HP:0002251, HP:0000175, S 2412	29430
${\rm HP:}0005807, {\rm HP:}0002335, {\rm HP:}0000126, {\rm HP:}0002334,$	
$\mathrm{HP:}0003155, \mathrm{HP:}0003037, \mathrm{HP:}0000316, \mathrm{HP:}0000455,$	
HP:0009748, HP:0009617, HP:0005781, HP:0001629	
PIGV Patient 7 HP:0004378, HP:0002251, HP:0002194, HP:0001655, S 2412	29430
${\rm HP:}0000175, {\rm HP:}0000126, {\rm HP:}0001195, {\rm HP:}0003155,$	
$\mathrm{HP:}0001250, \mathrm{HP:}0000455, \mathrm{HP:}0001252, \mathrm{HP:}0011344,$	
$\mathrm{HP:}0000750, \mathrm{HP:}0009836, \mathrm{HP:}0001344, \mathrm{HP:}0005484,$	
HP:0000271	
PIGV Patient 2 HP:0009882, HP:0002251, HP:0001821, HP:0003155, S 2412	29430
$\mathrm{HP:}0000316, \mathrm{HP:}0000455, \mathrm{HP:}0001252, \mathrm{HP:}0011344,$	
$\mathrm{HP:}0000750, \mathrm{HP:}0009836, \mathrm{HP:}0000431, \mathrm{HP:}0005484,$	
HP:0010804, HP:0000271, HP:0010819	
PIGV Patient 3 HP:0002251, HP:0006118, HP:0000077, HP:0003155, S 2412	29430
$\mathrm{HP:}0000316, \mathrm{HP:}0001250, \mathrm{HP:}0000455, \mathrm{HP:}0011398,$	
$\mathrm{HP:}0001252, \mathrm{HP:}0011344, \mathrm{HP:}0000750, \mathrm{HP:}0009836,$	
HP:0000431, HP:0010804, HP:0000271	
PIGV Patient 6 HP:0003155, HP:0000316, HP:0002251, HP:0002194, S 2412	29430
$\mathrm{HP:}0000455, \mathrm{HP:}0011398, \mathrm{HP:}0001263, \mathrm{HP:}0000750,$	
HP:0006895, HP:0000271, HP:0000081, HP:0002814	
PIGV Patient 4 HP:0002123, HP:0005490, HP:0001792, HP:0003155, S 2412	29430
$\mathrm{HP:}0000316, \mathrm{HP:}0000767, \mathrm{HP:}0001250, \mathrm{HP:}0000455,$	
$\mathrm{HP:}0001252, \mathrm{HP:}0011344, \mathrm{HP:}0000750, \mathrm{HP:}0000431,$	
HP:0009381, HP:0005914, HP:0010804, HP:0000271	
PIGV Patient 1 HP:0006118, HP:0002069, HP:0002019, HP:0001792, S 2231	5194
$\mathrm{HP:}0000637, \mathrm{HP:}0002714, \mathrm{HP:}0000426, \mathrm{HP:}0001270,$	
HP:0011398, HP:0000311, HP:0000256, HP:0010804,	
${\rm HP:}0001545, {\rm HP:}0009894, {\rm HP:}0000391, {\rm HP:}0004378,$	
$\mathrm{HP:}0002650, \mathrm{HP:}0000126, \mathrm{HP:}0003155, \mathrm{HP:}0011343,$	
HP:0000378, HP:0001520, HP:0001510, HP:0001344,	
HP:0000271, HP:0010819	

Table S1	_	Continued	from	previous	page
			1	P	1

Gene	Patient	HPOs		• •		Group	PMID
	ID					1	
PIGV	Patient 3	HP:0000072,	HP:0006118,	HP:0002883,	HP:0001195,	S	22315194
		HP:0001250,	HP:0000303,	HP:0011398,	HP:0011344,		
		HP:0010804,	HP:0100023,	HP:0200006,	HP:0000280,		
		HP:0004378,	HP:0010864,	HP:0000077,	HP:0000126,		
		HP:0005326,	HP:0000316,	HP:0000699,	HP:0003155,		
		HP:0001061,	HP:0011343,	HP:0001263,	HP:0000750,		
		HP:0001562.	HP:0010746. HF	P:0001510	,		
PIGV	Patient 2	HP:0002251.	HP:0002013.	HP:0001623.	HP:0002558.	S	22315194
		HP:0002714.	HP:0000218.	HP:0001270.	HP:0001250.		
		HP:0000256.	HP:0005484.	HP:0010804.	HP:0009894.		
		HP:0000391.	HP:0004378.	HP:0040262.	HP:0012374.		
		HP:0003270.	HP:0000126.	HP:0003155.	HP:0001263.		
		HP:0000378.	HP:0010946.	HP:0001520.	HP:0001510.		
		HP:0001344.	HP:0000271	,	,		
PIGV	Patient 4	HP:0003155.	HP:0000316.	HP:0001250.	HP:0002069.	S	22315194
110,1	1 0010110 1	HP:0011398.	HP:0001263.	HP:0000431.	HP:0001344.	~	
		HP:0010804.	HP:0001525. HF	2:0000280			
PIGW	Patient	HP·0001999	HP·0003155	HP·0000431	HP·0000023	S	24367057
110.11	1 actione	HP·0012310	HP-0002521 HF	P·0010804	111 .0000020,	S	21001001
PIGW	Patient 1	HP·0001270	HP·0001250	HP-0001252	HP·0002098	S	27626616
110.11	i actoric i	HP:0001662	HP-0002527 HF	P-0010296 HP-0	010819	S	21020010
PIGW	Patient 2	HP:0002607	HP-0001270	HP-0002123	HP-0001250	S	27626616
110,00	i aticiti 2	HP:0001263	HP-0001252 HF	2:0000158 HP:	010819	D	21020010
PICV	Family	HP:0006965	HP:0006528	HP-0002013	HP-000/387	S	26293662
1101	Δ_{-II-1}	HP:0006118	HP:0004691	HP:0002376	HP:0000038	5	20255002
	71-11-1	HP:0011047	HP:00040518	HP:0002070,	HP:0011027		
		HP:0025152	HP:0003155	HP:0006380	HP:0001250		
		HP:0001385	HP-0001510 HE	2.0031165 HP.(003236		
PICV	Family B-	HP:0001505,	HP-0001910, 111	HP-000750	HP-000486	S	26293662
1101	II_2	HP:0005484	HP-000718	111.0000750,	111 .0000400,	5	20233002
PICV	Family B	HP:0002104	HP:000718	HD-0001263	HD:0000750	S	26203662
1101	II_3	HP:0002154,	HP-0000752 HF	2001205, 2.0000271 HP.()000154	5	20233002
PICV	II-5 Family	HP:0000023	HP-0000232, III	HP:0010043	HD-000202	S	26203662
1101		HD:0011027	HD:0000510	HD.0002825	HD:0002027,	5	20293002
	A-11-2	HP:0002617	HP:0001250	HP.0002855, HP.0003273	HP:0001561		
		HP:0002017,	HP:0001510 HE	20003275,	111.0001501,		
	10	HD:0003020,	ир.0001010, 111 ир.0002122	.0005250 НР:0001979	HD-0001963	$\mathbf{T} + \mathbf{P}$	20100005
GIAAI	la	HP:0001310	HP.0002123, HP.0000038	HP:0025100	HP:0001203,	$1 \pm W$	29100095
		HP:0001240	HP-0011170 HE	0.0010810	111.0000033,		
CPA A1	1h	HP:000249,	HP:0001260	HP-0001979	HD:0001263	$T \perp P$	20100005
GIAAI	10	HD:0001210	HD:0001200,	HD.0001272,	HD:0001203,	ττι	29100095
		HD:0001310,	ни .0000958, нр.0010810	111.0025190,	111.0000039,		
	2.	ПП .0001249, ПР.0004299	ир.0000019	UD.0009464	UD.0000029	$\mathbf{T} + \mathbf{D}$	20100005
GFAAI	Ja	11F.0004522, 11D.0000620	11F.0002009, 11D.0001200	11F.0002404, 11D.0000216	IIF:0000938,	1+n	29100095
		HD.0000411	HP.0001290,	HD:0000510,	HP.0001272,		
		IIF:0000411,	IIF:0001203,	11F.0000520,	1117.0000341,		
	2 h	$\Pi P:0000232,$ $\Pi D:0002122$	ПР:0001249 П. 0001979	11D.0009060	IID.0000411	$\mathbf{T} + \mathbf{D}$	20100005
GPAAI	<u>ən</u>	ПР:0002125, ПР:0001262	HP:0001272, HP:0000445	HP:0002009, HD:0000520	ПР:0000411, ПР:0000028	1+n	29100095
		HD.0000620	HD.0001940 HT		111 .0000938,		
	40	IIF:0000039,	пг :0001249, ПГ пр.0004299	UUU1290	UD.000620	$\mathbf{T} + \mathbf{D}$	20100005
GLAU	4a	HD.0001979	HD.0001969	HD:0001957	HD.0000469	1+U	29100090
		HD.0000241	HD.0011990 HT	111.0001207,	111 .0000405,		
	4b	нг:0000341, пр.0000066	пг :0011220, ПГ пр.0004222	UUU1249	UD.0001979	$\mathbf{T} + \mathbf{D}$	20100005
GFAAI	40	ПГ:0002000, ПD:0001069	пг:0004322, пр.0001210	HD.0001957	11F:0001272,	1 + U	29100099
		пг:0001203, П. 0000241	пг:0001310, Пр.0011000 ПТ	nr:0001257,	пг:0000403,		
		нP:0000341,	пР:0011220, НЕ	10001249			

Gene	Patient ID	HPOs				Group	PMII
GPAA1	4c	HP:0002066,	HP:0004322,	HP:0000938,	HP:0001260,	T+R	2910009
		HP:0001272,	HP:0001263,	HP:0001310,	HP:0001257,		
		HP:0000463, I	HP:0011220, HI	P:0001249			
GPAA1	2	HP:0002069,	HP:0001263,	HP:0100704,	HP:0000938,	T+R	2910009
		HP:0000463,	HP:0000341,	HP:0011220,	HP:0001249,		
		HP:0001290	,	,	,		
GPAA1	$5\mathrm{b}$	HP:0002066,	HP:0001260,	HP:0002069,	HP:0001272,	T+R	2910009
		HP:0001263,	HP:0000445,	HP:0001310,	HP:0001249,		
		HP:0001290	,	,	,		
GPAA1	5a	HP:0002066,	HP:0001260,	HP:0002121,	HP:0002069,	T+R	2910009
		HP:0001272,	HP:0001263,	HP:0001310,	HP:0001249,		
		HP:0010819, I	HP:0001290	,	,		
PGAP1	Proband	HP:0002099,	HP:0001319,	HP:0001263,	HP:0000486,	T+R	2580440
		HP:0009748,	HP:0009904,	HP:0011968,	HP:0000395,		
		HP:0000582,	HP:0000639,	HP:0000490,	HP:0002099,		
		HP:0001319.	HP:0001263.	HP:0000486.	HP:0009748.		
		HP:0009904.	HP:0011968.	HP:0000395.	HP:0000582.		
		HP:0000639.	HP:0000490	,	, , ,		
PGAP1	1.2	HP:0000733,	HP:0008936,	HP:0001270,	HP:0004322,	T+R	2605093
		HP:0000556,	HP:0002791,	HP:0000248,	HP:0001263,		
		HP:0011344, I	HP:0000750, HI	P:0009062, HP:0)002090		
PGAP1	1.1	HP:0000193,	HP:0004322,	HP:0006934,	HP:0001508,	T+R	2605093
		HP:0001642,	HP:0001631,	HP:0010055,	HP:0000504,		
		HP:0000316,	HP:0000556,	HP:0000767,	HP:0001263,		
		HP:0001252.	HP:0000750.	HP:0000664.	HP:0001276.		
		HP:0001344.	HP:0005484	,	,		
PGAP1	II-1	HP:0006801.	HP:0012444.	HP:0002421.	HP:0002098.	T+R	2720673
-		HP:0000639.	HP:0002104.	HP:0001290.	HP:0001270.		
		HP:0003487.	HP:0000750.	HP:0025047.	HP:0011400.		
		HP:0005484.	HP:0000496, HI	P:0001336, HP:0	0001337		
PGAP1	II-2	HP:0012547.	HP:0006801.	HP:0012444.	HP:0002421.	T+R	2720673
		HP:0002098.	HP:0012101.	HP:0000639.	HP:0002104.	- 1 - 0	
		HP:0001290.	HP:0030874.	HP:0001270.	HP:0003487.		
		HP:0001250.	HP:0025047.	HP:0005484.	HP:0001336.		
		HP:0001337	,				
PGAP1	MR079	HP:0000733	HP:0002121	HP:0001319	HP:0001270	T+R	2478413
1		HP:0012444	HP:0000400	HP:0001263	HP:0010864	1 1V	21,0110
		HP:0000252		,			
PGAP1	Patient	HP:0012448	HP:0012447.	HP:0002079.	HP:0012443	T+R	2582341
	1 0010110	HP:0100704	HP:0011968	HP:0100660	HP:0011398	÷ , ±0	2002011
		HP:0001263	HP:0000750	HP:0010845	HP:0012429		
		IID.0000271	ID.0001940	,			

Table S1 Continued from previo

Table $\mathbf{S1}$ – Continued from previous page

Gene	Patient	HPOs	51 Continued	Jioni precious	page	Group	PMID
Gene	ID	111 05				Group	1 MIL
PGAP2	$\frac{12}{V.6/MR5}$	HP-0003155	HP-0001270	HP-0002360	HP-0004322	T+B	23561846
1 0111 2	v.0/11100	HP:0010864	HP:0001270,	HP:0000486	HP:0001324	1 10	20001010
		HP:0002059	HP:0001202,	HP:0002536	HP:0003155		
		HP:0001270	HP:0002360	HP:0004322	HP:0010864		
		$HP \cdot 0001270$, $HP \cdot 0001252$	HP:0000486	$HP \cdot 0001324$	HP:0002059		
		$HP \cdot 0001202,$	HP:0002536	HP $\cdot 0001524$,	HP:0001270		
		HP:0003202	HP:000/2320,	HP:0010864	HP:0001270,		
		HP:00013202,	HP:0004522,	$HP \cdot 0000004;$	HP:0001270		
		HP:0001324,	HP:0004322	HP:0010864	HP:0001270,		
		$HP \cdot 0003202,$	HP: 0004322 ,	$HP \cdot 0010304$	$HP \cdot 0000730$,		
		HP:0001232,	HP:0001324,	HP:0010864	HP:0002121,		
		$HP \cdot 0002526$	HP:0002300,	HP:00010004,	HP:0002000,		
		HP.0002550,	HP:0000750	HP:00001270,	HP:00013202,		
		HD.0001202,	HD:0000730,	HD:000430,	HD:0001324,		
		IIF .0001303, UD.0010864	HP:0002121,	ПГ:0004522, ПР:0002526	HP.0002300,		
		HP.00010804,	ПР:0002059, ПР:0002059	ПР:0002550, ПР:0001252	HP.0003133,		
		HP:0001270, HD:0000486	пР:0003202, ud.0001294 ит	$\Pi P:0001252,$	пР:0000750,		
DCAD9	TV.1	HP:0000480, I HD:0002155	IP:0001524, III	-:0001303 HD.0003260	UD.0004299	$\mathbf{T} + \mathbf{D}$	22561246
FGAF2	1V:1- MR042	HP.0010864	HP:0001270,	HP:0002300,	HP:0004322,	$1 \pm n$	23301040
	WII1043	UD.0002050	HD.0001202,	ПГ.0000480, ПР.0002526	UD.0001324,		
		HF.0002059, HD.0001270	ПР:0001303, ПР:0002260	ПР:0002550, ПР:0004255	IIF :0003133, UD:0010864		
		$\Pi P:0001270,$ $\Pi D:0001252$	$\Pi P:0002500,$ $\Pi D:0000486$	$\Pi P:0004522,$ $\Pi D:0001224$	$\Pi P:0010804,$ $\Pi D:0002050$		
		$\Pi P:0001202,$ $\Pi D:0001205$	$\Pi P:0000480,$	пР:0001524,	пР:0002059,		
DCADO	D. 4:	HP:0001305, J	HP:0002530	UD-00100F1	110.0009244	TID	06070440
PGAP2	Patient 1	HP:0009058,	HP:0002100,	HP:0010851,	HP:0002344,	1 + R	20879448
		HP:0002521,	HP:0030746,	HP:0000739,	HP:0005469,		
		HP:0003155,	HP:0001250,	HP:0000707,	HP:0001203,		
		HP:0001252,	HP:0001205,	HP:0003088,	HP:0000252,		
	T 1· · 1 1	HP:0002093, J	HP:0003324, HF	2:0011096, HP:0	JUU1946	T I D	00501047
PGAP2	Individual	HP:0003155,	HP:0001270,	HP:0001250,	HP:0000431,	1 + K	23561847
	A	HP:0010804, J	HP:0001249		UD 0000491	T I D	00501047
PGAP2	Individual	HP:0003155,	HP:0001270,	HP:0001250,	HP:0000431,	1 + R	23561847
	В	HP:0010804,	HP:0001249,	HP:0002251,	HP:0002079,		
		HP:0002025,	HP:0002650,	HP:0000175,	HP:0000154,		
		HP:0001631,	HP:0000637,	HP:0003155,	HP:0000316,		
		HP:0001270,	HP:0001250,	HP:0000455,	HP:0001252,		
		HP:0011344,	HP:0000750,	HP:0000431,	HP:0000365,		
		HP:0005484, J	HP:0010804, HF	2:0001249		T I D	07071400
PGAP2	Patient 2	HP:0001270,	HP:0001250,	HP:0001263,	HP:0000750,	1 + R	27871432
		HP:0001510,	HP:0000252,	HP:0001249,	HP:0001270,		
		HP:0001250,	HP:0001263,	HP:0000750,	HP:0001510,		
		HP:0000252,	HP:0001249,	HP:0003155,	HP:0001270,		
		HP:0001250,	HP:0001263,	HP:0000750,	HP:0001510,		
		HP:0000252,	HP:0001249,	HP:0001270,	HP:0001250,		
		HP:0001263,	HP:0000750,	HP:0001510,	HP:0000252,		
DGADO		HP:0001249				T I D	20110105
PGAP2	Patient 4-	HP:0003155,	HP:0001263,	HP:0000750,	HP:0001256,	T+R	29119105
DCADO	IV-7	HP:0000805		HD 0001049	HD 0000040	T I D	20110105
PGAP2	Patient 2-	HP:0003155,	HP:0001250,	HP:0001263,	HP:0002342,	T+R	29119105
DOADO	1V-5 D	HP:0031491,	HP:0000708, HI	2:0000718		THE P	00110105
PGAP2	Patient 3-	HP:0003155,	HP:0000750,	HP:0001575,	HP:0001256,	T+R	29119105
DCLES	1V-6	HP:0000716				m · F	00110105
PGAP2	Patient 1-	HP:0002373,	HP:0003155,	HP:0000750,	HP:0001575,	T+R	29119105
	1V-3	HP:0001256,	HP:0000716				

Gene	Patient	HPOs	51 Continued	Jioni precious	puye	Group	PMID
	ID						
PGAP3	Patient7-	HP:0002187,	HP:0002079,	HP:0002069,	HP:0002553,	T+R	28390064
	Family7	HP:0000175,	HP:0002265,	HP:0000023,	HP:0012450,		
		HP:0003194,	HP:0000028,	HP:0000717,	HP:0000637,		
		HP:0000448,	HP:0000316,	HP:0001270,	HP:0003155,		
		HP:0012390,	HP:0001250,	HP:0003763,	HP:0011398,		
		HP:0000431,	HP:0000752, HI	P:0005484, HP:0	0010804		
PGAP3	Patient6-	HP:0002079,	HP:0012714,	HP:0002553,	HP:0000175,	T+R	28390064
	Family6	HP:0010864,	HP:0002265,	HP:0008501,	HP:0002119,		
		HP:0003194,	HP:0000717,	HP:0000637,	HP:0000448,		
		HP:0003155,	HP:0000316,	HP:0001270,	HP:0011398,		
		HP:0000431,	HP:0000752,	HP:0001344,	HP:0005484,		
		HP:0010804, 1	HP:0007930				
PGAP3	Patient4-	HP:0002079,	HP:0002553,	HP:0000175,	HP:0010864,	T+R	28390064
	Family4	HP:0002265,	HP:0002119,	HP:0003194,	HP:0000717,		
		HP:0000637,	HP:0000448,	HP:0003155,	HP:0000316,		
		HP:0001270,	HP:0000587,	HP:0011398,	HP:0001321,		
		HP:0000431,	HP:0000752,	HP:0001344,	HP:0005484,		
		HP:0010804					
PGAP3	Patient5-	HP:0002187,	HP:0000060,	HP:0002123,	HP:0002553,	T+R	28390064
	Family5	HP:0000175,	HP:0002265,	HP:0012880,	HP:0003194,		
		HP:0000717,	HP:0000637,	HP:0000448,	HP:0003155,		
		HP:0000316,	HP:0001270,	HP:0000767,	HP:0011398,		
		HP:0000431, 1	HP:0001344, HF	P:0010804			
PGAP3	Patient8-	HP:0002187,	HP:0002079,	HP:0002123,	HP:0002553,	T+R	28390064
	Family7	HP:0002265,	HP:0002119,	HP:0003194,	HP:0000717,		
		HP:0030047,	HP:0000637,	HP:0004325,	HP:0000448,		
		HP:0000316,	HP:0001270,	HP:0003155,	HP:0003763,		
		HP:0011398,	HP:0000431,	HP:0000752,	HP:0005484,		
50150	D	HP:0001344,	HP:0010804, HI	P:0001357, HP:0	001305	-	
PGAP3	Patient9-	HP:0002187,	HP:0002079,	HP:0002069,	HP:0002553,	T+R	28390064
	Family8	HP:0000175,	HP:0002265,	HP:0003194,	HP:0000028,		
		HP:0000717,	HP:0000637,	HP:0000448,	HP:0003155,		
		HP:0000316,	HP:0001270,	HP:0003763,	HP:0011398,		
		HP:0000431,	HP:0000752,	HP:0001344,	HP:0005484,		
DGIDA	D II II	HP:0000540,	HP:0010804			T D	2222222
PGAP3	Patient1-	HP:0002079,	HP:0002553,	HP:0010864,	HP:0002265,	T+R	28390064
	Family1	HP:0003194,	HP:0000639,	HP:0000717,	HP:0000637,		
		HP:0000448,	HP:0003155,	HP:0000316,	HP:0001270,		
		HP:0000204,	HP:0011398,	HP:0000202,	HP:0001321,		
		HP:0000431,	HP:0001344,	HP:0005484,	HP:0001433,		
DGADO	D	HP:0010804				T D	20200000
PGAP3	Patient10-	HP:0002079,	HP:0001601,	HP:0002553,	HP:0000175,	T+R	28390064
	Family8	HP:0010864,	HP:0002265,	HP:0000023,	HP:0003194,		
		HP:0000717,	HP:0000637,	HP:0000448,	HP:0003155,		
		HP:0000316,	HP:0001270,	HP:0011398,	HP:0000431,		
		HP:0000752,	HP:0001256,	нР:0001344,	HP:0000540,		
DOADO		HP:0010804					00000004
PGAP3	Patient2-	HP:0002187,	HP:0002079,	HP:0002553,	HP:0000175,	1 + K	28390064
	Family2	HP:0001643,	HP:0002265,	HP:0003194,	HP:0000717,		
		HP:000057,	пР:0000448,	HP:0000316,	HP:0001270,		
		HP:0003155,	HP:0000421	HP:0003763,	HP:0011398,		
		HP:0000202,	нг:0000431,	HP:0000752,	HP:0005484,		
		пР:0001344,	пг:0002092, HI	-:0010804, HP:(010808		

Table **S1** – Continued from previous page

10 11 11+1 28390064 PGAP3 Patient3- Family3 HP-0002079, HP-0001260, HP-0001386, HP-0001270, HP-0001280, HP-0001386, HP-0002187, HP-0001380, HP-0000136, HP-00002185, HP-0000431, HP-0000316, HP-0000315, HP-0000772, HP-0000316, HP-0000315, HP-0000772, HP-00001305, T+R T+R 24439110 PGAP3 V-2- Family HP-00002069, HP-0000175, HP-00002265, HP-0000316, HP-0000270, HP-00002185, HP-0000750, HP-0000265, HP-0000270, HP-0000270, HP-0000265, HP-0000270, HP-0000270, HP-0000265, HP-0000270, HP-0000218, HP-0000266, HP-0000276, HP-0000218, HP-0000266, HP-0000276, HP-0000218, HP-0000218, HP-0000355, HP-0000218, HP-0000218, HP-0000455, HP-0000218, HP-0000218, HP-0000455, HP-0000218, HP-0000218, HP-0000455, HP-0000218, HP-0000218, HP-0000455, HP-0000218, HP-0000218, HP-0000455, HP-0000218, HP-0000218, HP-0000455, HP-0000218, HP-0000126, HP-0000455, HP-0000218, HP-0000126, HP-0000455, HP-0000218, HP-0000126, HP-0000455, HP-0001263, HP-0000226, HP-0000435, HP-0001261, HP-0000435, HP-0001263, HP-00002120, HP-0000435, HP-0001261, HP-0000435, HP-00002120, HP-0000435, HP-0000436, HP-0001263, HP-0000432, HP-0001263, HP-0000436, HP-0001263, HP-0000431, HP-0000458, HP-0000431, HP-0000458, HP-0004	Gene	Patient	HPOs		Jiem preese as	page	Group	PMID
PGAP3 Patents HP-0002205, HP-0001304, HP-0000285, HP-00003163, HP-00003164, HP-0000544, HP-0000540, HP-0001308, HP-00003165, HP-00003165, HP-00003155, HP-00003155, HP-0000316, HP-00003155, HP-00003155, HP-00003155, HP-00003155, HP-00003155, HP-00003155, HP-00003155, HP-00003156, HP-00002763, HP-00001283, HP-0000278, HP-00003156, HP-0000315, HP-00003156, HP-0000316, HP-0001226, HP-0000316, HP-0001226, HP-0000316, HP-0001236, HP-0000316, HP-0001236, HP-0000316, HP-0001236, HP-00001236, HP-0001236, HP-00001236, HP-0001236, HP-00001236	DGLDO	ID		TID 0000000				202000001
Framity3 HP-0000430, HP-0001270, HP-0001270, HP-0001316, HP-0002553, HP-0000752, HP-0000752, HP-0000136, HP-0002553, HP-0000752, HP-0000136, HP-0002185, HP-0000175, HP-0002265, HP-00001270, A T+R 24439110 PGAP3 V-2 HP-0002069, HP-0000175, HP-0002265, HP-0002186, HP-0002120, HP-0000155, HP-0000236, HP-0002263, HP-0000156, HP-0000582, HP-0000236, HP-0002265, HP-00002548, HP-00010544, HP-00010544, HP-00010544, HP-0000582, HP-0002270, HP-0002365, HP-0000582, HP-00001816, HP-0001270, HP-0000570, HP-0000455, HP-0001270, HP-0001364, HP-0001250, HP-0001270, HP-0001316, HP-000155, HP-0000256, HP-0001264, HP-0000456, HP-0001270, HP-0001364, HP-0000750, HP-0000750, HP-0000316, HP-0001270, HP-0001316, HP-0010804 HP-0000750, HP-0000582, HP-0000316, HP-0001261, HP-0001365, HP-0001270, HP-0000455, HP-0001270, HP-0001316, HP-0001270, HP-0000451, HP-0000582, HP-000120, HP-0000431, HP-0000453, HP-0001270, HP-0000452, HP-0001316, HP-0001270, HP-0000431, HP-0001220, HP-0001316, HP-0001270, HP-0000431, HP-0001220, HP-0001316, HP-0001363, HP-0001270, HP-0000451, HP-0000582, HP-0001220, HP-0001316, HP-0001370, HP-0000431, HP-0001220, HP-0000316, HP-0000316, HP-0000316, HP-0001220, HP-0000316, HP-0000316, HP-0001314, HP-0000582, HP-0001314, HP-0000582, HP-0000316, HP-0000316, HP-0001370, HP-0000316, HP-0000316, HP-0001370, HP-0000316, HP-0000316, HP-0001370, HP-0000316, HP-0001270, HP-0001370, HP-0000316, HP-0001270, HP-0001370, HP-0000314, HP-0001283, HP-0001370, HP-0000314, HP-0001283, HP-0001370, HP-0000314, HP-0001283, HP-0001384, HP-0001283, HP-0001284, HP-0001384, HP-0000314, HP-0001283, HP-0001384, HP-0000315, HP-0000313, HP-0001283, HP-0001384, HP-0000315, HP-0000313, HP-0001283, HP-0001384, HP-0000315, HP-0000313, HP-0001283, HP-0001384, HP-0000315, HP-0000314, HP-0000384, HP-0000315, HP-0000314, HP-0000384, HP-0000315, HP-0000314, HP-00000314, HP-0000284, HP-0000314, HP-0000314, HP-0000314, HP-000031	PGAP3	Patient3-	HP:0002079,	HP:0002069,	HP:0002265,	HP:0001631,	T+R	28390064
PGAP3 V-2. PP:0001344, PP:0000316, PP:0000316, PP:0000316, PP:0000315, PP:0000316, PP:0000268, PP:0000268, PP:0000268, PP:0000270, PP:0000316, PP:00000316, PP:0000316, PP:0000316, PP:0000316, PP:0000316, PP:00003		Family3	HP:0008501,	HP:0003194,	HP:0000637,	HP:0000448,		
PGAP3 V-2. IP-0002553, IP-0006872, IP-0000752, IP-0001433, IP-0001343, IP-0001343, IP-0001343, IP-0001355, IP-0001354, IP-0001364, IP-0001364, IP-0001376, IP-0000316, IP-0000316, IP-0001270, IP-0000316, IP-0000316, IP-0000263, IP-0000263, IP-0000263, IP-000028, IP-000028, IP-0000270, IP-000028, IP-000028, IP-0000270, IP-000028, IP-0000270, IP-0000276, IP-0000136, IP-0001376, IP-0001376, IP-0001376, IP-0001270, IP-0000136, IP-0001376, IP-0001270, IP-0000437, IP-0000276, IP			HP:0001270,	HP:0001250,	HP:0011398,	HP:0003186,		
HP-20002363, HP-2000135, HP-2000717, HP-2000133, HP-2000135, HP-2000218, HP-2000175, HP-2000225, HP-2000236, A HP-2000218, HP-2000175, HP-2002255, HP-20002170, A HP-2000218, HP-2000125, HP-20002184, HP-20001220, HP-20002123, HP-20002180, HP-2002265, T+R 24439110 PGAP3 II-1 HP-20002152, HP-20002134, HP-20002184, HP-2001284, HP-2000235, HP-20002123, HP-20002126, HP-20002316, C HP-20002182, HP-20012810, HP-2002265, T+R 24439110 PGAP3 V-3. HP-20002152, HP-20002126, HP-20002186, HP-20002316, HP-20000431, HP-2000279, IP-20001264, HP-20002185, HP-20000316, IP-20002128, HP-2000218, HP-20002185, HP-20002763, HP-20002128, HP-2000218, HP-2000264, HP-20000582 T+R 24439110 PGAP3 V-3. HP-20002123, HP-20002126, HP-2000218, HP-20002170, HP-20002763, IP-20002123, HP-20002164, HP-2000184, HP-20001250, HP-20002126, HP-2000164, HP-2001270, HP-20001250, HP-2000316, HP-20001263, HP-2001220, HP-2000316, HP-20001316, HP-2001270, HP-20001314, HP-20001864, HP-20001270, HP-20001314, HP-2000186, HP-20002184, HP-2001220, HP-20001316, HP-20001270, HP-20001316, HP-20001316, HP-20001270, HP-20001316, HP-20001251, HP-20003155, HP-20001316, HP-20002126, HP-20003155, HP-20001316, HP-20002126, HP-20003155, HP-20001316, HP-20002127, HP-20002180, HP-2000150, HP-2000150, HP-20003155, HP-2000150, HP-2000150, HP-20003155, HP-2000150, HP-2000150, HP-20003155, HP-2000150, HP-2000150, HP-20003155, HP-2000150, HP-2000150, HP-20003155, HP-2000150, HP-2000150, HP-20003155, HP-2000150, HP-2000150, HP-2000150, HP-2000150, HP-2000150, HP-2000150, HP-2000150, HP-2000150, HP-2000150, HP-2000150, HP-2000150, HP-20001525, HP-2000150, HP-2000150, HP-2000150, HP-2000150, HP-2000150, HP-2000150, HP-2000150, HP-2000150, HP-2000150, HP-2000150, HP-2000150, HP-2000150, HP-2			HP:0005484,	HP:0010804,	HP:0010808,	HP:0002187,		
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PGAP3 V-2- Family HP:000266, HP:000315, HP:000315, HP:000315, HP:000315, HP:000315, HP:000315, HP:000316, HP:000263, HP:000263, HP:000263, HP:000263, HP:000275, HP:0000431, HP:000275, HP:0000431, HP:000275, HP:0000276, HP:00			HP:0003155,	HP:0000431,	HP:0000752,	HP:0001433,		
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HP:0000431, HP:0000365, HP:0001344, HP:0010804, HP:0000582, HP:0001629, HP:0000490			HP:0000733	HP:0001270	HP:0001263	HP:0009748		
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			HP:0000582.	HP:0001629. HF	P:0000490			

Gene	Patient	HPOs	Si continued	Jient preetoue	page	Group	PMID
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PGAP3	257982	HP:0012810,	HP:0008935,	HP:0006108,	HP:0002194,	T+R	28327575
		HP:0000175,	HP:0012345,	HP:0025335,	HP:0002136,		
		HP:0000219,	HP:0000369,	HP:0004619,	HP:0003186,		
		HP:0000286,	HP:0001488,	HP:0001357,	HP:0001305,		
		HP:0010818,	HP:0002121,	HP:0002751,	HP:0000154,		
		HP:0001763,	HP:0000319,	HP:0000316,	HP:0100543,		
		HP:0003155,	HP:0002938,	HP:0000403,	HP:0000687,		
		HP:0001344					
PGAP3	257982 -	HP:0012810,	HP:0008935,	HP:0006108,	HP:0002194,	T+R	28327575
	affected	HP:0000175,	HP:0012345,	HP:0025335,	HP:0002136,		
	brother	HP:0000219,	HP:0000369,	HP:0004619,	HP:0003186,		
		HP:0000286,	HP:0001488,	HP:0001357,	HP:0001305,		
		HP:0010818,	HP:0002121,	HP:0000154,	HP:0001763,		
		HP:0000319,	HP:0000316,	HP:0100543,	HP:0003155,		
		HP:0002938,	HP:0000403, HI	P:0000687, HP:0	001344		
PGAP3	proband	HP:0001769,	HP:0002079,	HP:0002360,	HP:0006118,	T+R	29531774
		HP:0002650,	HP:0000175,	HP:0002197,	HP:0000028,		
		HP:0000736,	HP:0001290,	HP:0003155,	HP:0011443,		
		HP:0000369,	HP:0000347,	HP:0003763,	HP:0001263,		
		HP:0000278,	HP:0000431, HI	P:0010804, HP:0	001837		
PIGT	Patient	HP:0002384,	HP:0000072,	HP:0006429,	HP:0000121,	T+R	23636107
	V-2	HP:0002750,	HP:0000938,	HP:0000113,	HP:0001290,		
		HP:0005622,	HP:0003100,	HP:0000248,	HP:0000256,		
		HP:0000496,	HP:0009824,	HP:0000472,	HP:0004443,		
		HP:0002650,	HP:0010852,	HP:0003282,	HP:0025330,		
		HP:0000505,	HP:0002648,	HP:0003022,	HP:0001263,		
		HP:0006480,	HP:0001520,	HP:0000540,	HP:0001249,		
		HP:0000271,	HP:0100864				
PIGT	Patient	HP:0006429,	HP:0000121,	HP:0002750,	HP:0000938,	T+R	23636107
	V-1	HP:0011648,	HP:0000639,	HP:0004349,	HP:0001290,		
		HP:0000767,	HP:0000248,	HP:0000486,	HP:0000256,		
		HP:0009824,	HP:0000496,	HP:0011330,	HP:0002123,		
		HP:0002650,	HP:0010852,	HP:0100704,	HP:0000505,		
		HP:0002648,	HP:0001263,	HP:0003022,	HP:0006480,		
		HP:0001520,	HP:0000540, HI	P:0000271, HP:0	001249		
PIGT	Patient	HP:0000072,	HP:0002283,	HP:0002069,	HP:0000121,	T+R	23636107
	V-5	HP:0100704,	HP:0002750,	HP:0004349,	HP:0000639,		
		HP:0001272,	HP:0000486,	HP:0006480,	HP:0003186,		
		HP:0009824,	HP:0000540, HI	P:0003429, HP:0	0001249		
PIGT	Patient	HP:0007360,	HP:0006429,	HP:0000072,	HP:0002283,	T+R	23636107
	V-4	HP:0000121,	HP:0002750,	HP:0000938,	HP:0001290,		
		HP:0005622,	HP:0003100,	HP:0000248,	HP:0000256,		
		HP:0003186,	HP:0009824,	HP:0000496,	HP:0000472,		
		HP:0001638,	HP:0002123,	HP:0012697,	HP:0003282,		
		HP:0000505,	HP:0002648,	HP:0001263,	HP:0003022,		
		HP:0006480,	HP:0001520,	HP:0000540,	HP:0000271,		
		HP:0100864,	HP:0001249				

Gene	Patient ID	HPOs				Group	PMID
PIGT	Patient 2	HP:0009127, HP:0	0005280,	HP:0002069,	HP:0010841,	T+R	25943031
		HP:0025458, HP:0	0001631,	HP:0002720,	HP:0000938,		
		HP:0030718, HP:0	0002334,	HP:0002059,	HP:0000639,		
		HP:0000924, HP:0	0002835,	HP:0002714,	HP:0002705,		
		HP:0000545, HP:0	0003100,	HP:0011994,	HP:0000248,		
		HP:0001250, HP:0	0001272,	HP:0000767,	HP:0011996,		
		HP:0000486, HP:0	0000343,	HP:0000365,	HP:0000341,		
		HP:0001513, HP:0	0009131,	HP:0001337,	HP:0001537,		
		HP:0002187, HP:0	0002650,	HP:0002870,	HP:0002020,		
		HP:0002850, HP:0	0100704,	HP:0030856,	HP:0030854,		
		HP:0000348, HP:0	0001382,	HP:0001263,	HP:0000463,		
		HP:0001336, HP:001	10819				
PIGT	Patient 1	HP:0009127, HP:0	0005280,	HP:0001657,	HP:0002069,	T+R	25943031
		HP:0010841, HP:0	0025458,	HP:0000938,	HP:0002334,		
		HP:0002059, HP:0	0000639,	HP:0002835,	HP:0002714,		
		HP:0002705, HP:0	0003100,	HP:0001272,	HP:0000248,		
		HP:0000486, HP:0	0000365,	HP:0000343,	HP:0000484,		
		HP:0000341, HP:0	0001929,	HP:0005616,	HP:0001513,		
		HP:0009131, HP:0	0003225,	HP:0002155,	HP:0002650,		
		HP:0002020, HP:0	0100704,	HP:0006254,	HP:0000826,		
		HP:0000348, HP:00	11167, HF	P:0000463, HP:0	0001336		
PIGT	female	HP:0000072, HP:0	0005280,	HP:0011648,	HP:0000639,	T+R	24906948
	proband	HP:0002059, HP:0	0008872,	HP:0002714,	HP:0000218,		
		HP:0000787, HP:0	0001250,	HP:0000347,	HP:0000369,		
		HP:0001272, HP:0	0001252,	HP:0001561,	HP:0000486,		
		HP:0010804, HP:0	0000272,	HP:0007366,	HP:0002187,		
		HP:0000071, HP:0	0002133,	HP:0006913,	HP:0002650,		
		HP:0002119, HP:0	0000939,	HP:0003282,	HP:0000505,		
		HP:0000463, HP:00	00582				
PIGT	258094	HP:0002373, HP:0	0006855,	HP:0002069,	HP:0002073,	T+R	28327575
		HP:0002465, HP:0	0025331,	HP:0000505,	HP:0001260,		
		HP:0001270, HP:0	0000787,	HP:0001182,	HP:0001251,		
		HP:0001272, HP:0	0001250,	HP:0000657,	HP:0000455,		
		HP:0001263, HP:00	00490				
PIGT	270250	HP:0002187, HP:0	0040288,	HP:0007141,	HP:0001250,	T+R	28327575
		HP:0001283, HP:3	3000033,	HP:0200134,	HP:0002353,		
		HP:0000252, HP:00	03282, HF	P:0100765, HP:0	0011471		
PIGT	270306	HP:0002187, HP:0	0040288,	HP:0007141,	HP:0001250,	T+R	28327575
		HP:0001283, HP:3	3000033,	HP:0200134,	HP:0002353,		
		HP:0000252, HP:00	03282. HF	P:0100765, HP:0	0011471		

Protein	Symbol
Acetylcholinesterase (AChE) (EC 3.1.1.7)	ACHE
Intestinal-type alkaline phosphatase (IAP) (Intestinal alkaline phosphatase) (EC 3.1.3.1)	ALPI
Alkaline phosphatase, tissue-nonspecific isozyme (AP-TNAP) (TNSALP) (EC 3.1.3.1) (Alkaline phosphatase liver/bone/kidney isozyme)	ALPL
Alkaline phosphatase, placental type (EC 3.1.3.1) (Alkaline phosphatase Regan isozyme) (Placental alkaline phosphatase 1) (PLAP-1)	ALPP
Alkaline phosphatase, placental-like (EC 3.1.3.1) (ALP-1) (Alkaline phosphatase Nagao isozyme) (Germ cell alkaline phosphatase) (GCAP) (Placental alkaline phosphatase-like) (PLAP-like)	ALPPL2
GPI-linked NAD(P)(+)-arginine ADP-ribosyltransferase 1 (EC 2.4.2.31) (ADP-ribosyltransferase C2 and C3 toxin-like 1) (ARTC1) (Mono(ADP-ribosyl)transferase 1) (CD antigen CD296)	ART1
Ecto-ADP-ribosyltransferase 3 (EC 2.4.2.31) (ADP-ribosyltransferase C2 and C3 toxin-like 3) (ARTC3) (Mono(ADP-ribosyl)transferase 3) (NAD(P)(+)-arginine ADP-ribosyltransferase 3)	ART3
Ecto-ADP-ribosyltransferase 4 (EC 2.4.2.31) (ADP-ribosyltransferase C2 and C3 toxin-like 4) (ARTC4) (Dombrock blood group carrier molecule) (Mono(ADP-ribosyl)transferase 4) (NAD(P)(+)-arginine ADP-ribosyltransferase 4) (CD antigen CD297)	ART4
Brevican core protein (Brain-enriched hyaluronan-binding protein) (BEHAB) (Chon- droitin sulfate proteoglycan 7)	BCAN
ADP-ribosyl cyclase/cyclic ADP-ribose hydrolase 2 (EC 3.2.2.6) (ADP-ribosyl cyclase 2) (Bone marrow stromal antigen 1) (BST-1) (Cyclic ADP-ribose hydrolase 2) (cADPr hydrolase 2) (CD antigen CD157)	BST1
Bone marrow stromal antigen 2 (BST-2) (HM1.24 antigen) (Tetherin) (CD antigen CD317)	BST2
Carbonic anhydrase 4 (EC 4.2.1.1) (Carbonate dehydratase IV) (Carbonic anhydrase IV) (CA-IV)	CA4
Calcium channel, voltage-dependent, alpha-2/delta subunit 2 CD109 antigen (150 kDa TGF-beta-1-binding protein) (C3 and PZP-like alpha-2- macroglobulin domain-containing protein 7) (Platelet-specific Gov antigen) (p180)	CACNA2D2 CD109
Monocyte differentiation antigen CD14 (Myeloid cell-specific leucine-rich glycopro- tein) (CD antigen CD14) [Cleaved into: Monocyte differentiation antigen CD14, uri- nary form: Monocyte differentiation antigen CD14, membrane-bound form]	CD14
CD160 antigen (Natural killer cell receptor BY55) (CD antigen CD160)	CD160
CD177 antigen (Human neutrophil alloantigen 2a) (HNA-2a) (NB1 glycoprotein) (NB1 GP) (Polycythemia rubra vera protein 1) (PRV-1) (CD antigen CD177)	CD177
Signal transducer CD24 (Small cell lung carcinoma cluster 4 antigen) (CD antigen CD24)	CD24
CD48 antigen (B-lymphocyte activation marker BLAST-1) (BCM1 surface antigen) (Leukocyte antigen MEM-102) (SLAM family member 2) (SLAMF2) (Signaling lymphocytic activation molecule 2) (TCT.1) (CD antigen CD48)	CD48
CAMPATH-1 antigen (CDw52) (Cambridge pathology 1 antigen) (Epididymal secre- tory protein E5) (Human epididymis-specific protein 5) (He5) (CD antigen CD52)	CD52
Complement decay-accelerating factor (CD antigen CD55)	CD55
Cd58, LFA3, lymphocyte function-associated antigen, type 3	CD58
CD59 glycoprotein (1F5 antigen) (20 kDa homologous restriction factor) (HRF-20) (HRF20) (MAC-inhibitory protein) (MAC-IP) (MEM43 antigen) (Membrane attack complex inhibition factor) (MACIF) (Membrane inhibitor of reactive lysis) (MIRL) (Protectin) (CD antigen CD59)	CD59
Cadherin-13 (Heart cadherin) (H-cadherin) (P105) (Truncated cadherin) (T-cad) (T-cadherin)	CDH13
Carcinoembryonic antigen-related cell adhesion molecule 5 (Carcinoembryonic anti- gen) (CEA) (Meconium antigen 100) (CD antigen CD66e)	CEACAM5

Table 52 Continued from previous page	
Protein	Symbol
Carcinoembryonic antigen-related cell adhesion molecule 6 (Non-specific crossreacting	CEACAM6
antigen) (Normal cross-reacting antigen) (CD antigen CD66c)	
Carcinoembryonic antigen-related cell adhesion molecule 7 (Carcinoembryonic antigen	CEACAM7
CGM2)	
Carcinoembryonic antigen-related cell adhesion molecule 8 (CD67 antigen) (Carci-	CEACAM8
noembryonic antigen CGM6) (Non-specific cross-reacting antigen NCA-95) (CD anti-	
gen CD66b)	
Cryptic protein (Cryptic family protein 1)	CFC1
Ciliary neurotrophic factor receptor subunit alpha (CNTF receptor subunit alpha)	CNTFR
(CNTFR-alpha)	ONTIN
Contactin-1 (Chycoprotein gn135) (Neural cell surface protein F3)	CNTN1
Contactin 2 (Avonal glycoprotein TAC 1) (Avonal 1) (Transient avonal glycoprotein	CNTN2
(1) (TAV 1)	011112
1) (IAA-1) Contactin 2 (Drain derived immunoglobulin superfemily protein 1) (DIC 1)	CNTN2
(Discussed and the control of the co	UNTINS
(Plasmacytoma-associated neuronal glycoprotein)	CINITENI (
Contactin-4 (Brain-derived immunoglobulin superfamily protein 2) (BIG-2)	CNTN4
Contactin-5 (Neural recognition molecule NB-2) (hNB-2)	CNTN5
Contactin-6 (Neural recognition molecule NB-3) (hNB-3)	CNTN6
Carboxypeptidase M (CPM) (EC 3.4.17.12)	CPM
Carboxypeptidase O (CPO) (EC 3.4.17)	CPO
Dipeptidase (EC $3.4.13.19$)	DPEP1
Dipeptidase (EC 3.4.13.19) (Fragment)	DPEP2
Dipeptidase 3 (EC $3.4.13.19$)	DPEP3
Ephrin-A1 (EPH-related receptor tyrosine kinase ligand 1) (LERK-1) (Immediate	EFNA1
early response protein B61) (Tumor necrosis factor alpha-induced protein 4) (TNF	
alpha-induced protein 4) [Cleaved into: Ephrin-A1, secreted form]	
Ephrin-A2 (EPH-related receptor tyrosine kinase ligand 6) (LERK-6) (HEK7 ligand)	EFNA2
(HEK7-L)	
Ephrin-A3 (EFL-2) (EHK1 ligand) (EHK1-L) (EPH-related receptor tyrosine kinase	EFNA3
ligand 3) (LERK-3)	
Ephrin-A4 (EPH-related receptor tyrosine kinase ligand 4) (LERK-4)	EFNA4
Ephrin-A5 (AL-1) (EPH-related receptor tyrosine kinase ligand 7) (LEBK-7)	EFNA5
Ectonucleotide pyrophosphatase/phosphodiesterase family member 6 (E-NPP 6)	ENPP6
(NPP-6) (EC 3 1 4 -) (EC 3 1 4 38) (Choline-specific glycerophosphodiester phospho-	
diesterase) (Clycerophosphocholine cholinenhosphodiesterase) (CPC Cpde)	
Low affinity immunoglobulin gamma Fa region recentor III B (Fa gamma BIII bota)	FCCP3B
(Fa gamma DIII) (Fa gamma DIIIb) (FaDIII) (FaDIIIb) (FaDIIIb) (FaDIIIb)	rognab
(FC-gamma KIII) (FC-gamma KIIID) (FCKIII) (FCKIIID) (FCK-10) (IgG FC receptor	
III-1) (CD antigen CD100) E-late meanter alche (ED alche) (Adult falate binding metain) (EDD) (Ealate m	EOLD1
Folate receptor alpha (FR-alpha) (Adult folate-binding protein) (FBP) (Folate re-	FOLKI
ceptor 1) (Folate receptor, adult) (KB cells FBP) (Ovarian tumor-associated antigen	
MOv18)	DOLDO
Folate receptor beta (FR-beta) (Folate receptor 2) (Folate receptor, fetal/placental)	FOLR2
(Placental folate-binding protein) (FBP)	
Folate receptor gamma (FR-gamma) (Folate receptor 3)	FOLR3
Growth arrest-specific protein 1 (GAS-1)	GAS1
GDNF family receptor alpha-1 (GDNF receptor alpha-1) (GDNFR-alpha-1) (GFR-	GFRA1
alpha-1) (RET ligand 1) (TGF-beta-related neurotrophic factor receptor 1)	
GDNF family receptor alpha-2 (GDNF receptor alpha-2) (GDNFR-alpha-2) (GFR-	GFRA2
alpha-2) (GDNF receptor beta) (GDNFR-beta) (Neurturin receptor alpha) (NRTNR-	
alpha) (NTNR-alpha) (RET ligand 2) (TGF-beta-related neurotrophic factor receptor	
2)	
GDNF family receptor alpha-3 (GDNF receptor alpha-3) (GDNFR-alpha-3) (GFR-	GFRA3
alpha-3)	
GDNF family receptor alpha-4 (GDNF receptor alpha-4) (GDNFR-alpha-4) (GFR-	GFRA4
alpha-4) (Persephin receptor)	
GLIPR1-like protein 1	GLIPB1L1
Glycosyl-phosphatidylinositol-anchored molecule-like protein	GML
Gijoosji prosprandymosnor anchored molecule-nke protein	OINTE

Protein	Symbol
Pancreatic secretory granule membrane major glycoprotein GP2 (Pancreatic zymogen	GP2
granule membrane protein GP-2) (ZAP75)	
Glypican-1 [Cleaved into: Secreted glypican-1]	GPC1
Glypican-2 [Cleaved into: Secreted glypican-2]	GPC2
Glypican-3 (Fragment)	GPC3
K-glypican	GPC4
Glypican-5 [Cleaved into: Secreted glypican-5]	GPC5
Glypican-6 [Cleaved into: Secreted glypican-6]	GPC6
Glycosylphosphatidylinositol-anchored high density lipoprotein-binding protein 1 (GPI-HBP1) (GPI-anchored HDL-binding protein 1) (High density lipoprotein-	GPIHBP1
binding protein 1)	
Hemojuvelin (Hemochromatosis type 2 protein) (RGM domain family member C)	HFE2
noma protein 2) (LuCa-2)	HYAL2
Immunoglobulin superfamily member 21 (IgSF21)	IGSF21
Intelectin-1 (ITLN-1) (Endothelial lectin HL-1) (Galactofuranose-binding lectin) (In-	ITLN1
testinal lactoferrin receptor) (Omentin)	
Sperm-egg fusion protein Juno (Folate receptor 4) (Folate receptor delta) (FR-delta) (IZUMO1 receptor protein JUNO)	IZUMO1R
Lipoprotein lipase (LPL) (EC 3.1.1.34)	LPL
Limbic system-associated membrane protein (LSAMP) (IgLON family member 3)	LSAMP
Lymphocyte antigen 6D (Ly-6D) (E48 antigen)	LY6D
Lymphocyte antigen 6E (Ly-6E) (Retinoic acid-induced gene E protein) (RIG-E) (Stem cell antigen 2) (SCA-2) (Thymic shared antigen 1) (TSA-1)	LY6E
Lymphocyte antigen 6 complex locus protein G6c	LY6G6C
Lymphocyte antigen 6 complex locus protein G6d (Protein Ly6-D) (Megakaryocyte-	LY6G6D
enhanced gene transcript 1 protein)	
Lymphocyte antigen 6H (Ly-6H)	LY6H
Lymphocyte antigen 6K (Ly-6K)	LY6K
Lymphocyte antigen 6L (Lymphocyte antigen 6 complex locus protein L)	LY6L
Ly-6/neurotoxin-like protein 1 (Endogenous prototoxin LYNX1) (Testicular tissue	LYNX1
protein Li 112)	
Ly6/PLAUR domain-containing protein 1 (Putative HeLa tumor suppressor) (PHTS)	LYPDI
Lyb/PLAUR domain-containing protein 2	LYPD2
Ly6/PLAUR domain-containing protein 3 (GPI-anchored metastasis-associated pro- tein C4.4A homolog) (Matrigel-induced gene C4 protein) (MIG-C4)	LYPD3
Ly6/PLAUR domain-containing protein 4	LYPD4
Ly6/PLAUR domain-containing protein 5	LYPD5
Ly6/PLAUR domain-containing protein 6	LYPD6
Ly6/PLAUR domain-containing protein 6B	LYPD6B
Ly6/PLAUR domain-containing protein 8	LYPD8
MAM domain-containing glycosylphosphatidylinositol anchor protein 1 (Fragment)	MDGA1
MAM domain-containing glycosylphosphatidylinositol anchor protein 2	MDGA2
Melanotransferrin (Melanoma-associated antigen p97) (CD antigen CD228)	MELTF
Matrix metalloproteinase-17 (MMP-17) (EC 3.4.24) (Membrane-type matrix met-	MMP17
alloproteinase 4) (MT-MMP 4) (MTMMP4) (Membrane-type-4 matrix metallopro- teinase) (MT4-MMP) (MT4MMP)	
Matrix metalloproteinase-25 (MMP-25) (EC 3.4.24) (Leukolysin) (Membrane-type matrix metalloproteinase 6) (MT-MMP 6) (MTMMP6) (Membrane-type-6 matrix	$\rm MMP25$
metalloproteinase) (MT6-MMP) (MT6MMP)	
Mesothelin (CAK1 antigen) (Pre-pro-megakarvocyte-potentiating factor) [Cleaved	MSLN
nto: Megakarvocyte-potentiating factor (MPF). Mesothelin cleaved form]	11101211
Neural cell adhesion molecule 1 (N-CAM-1) (NCAM-1) (CD antigen CD56)	NCAM1
Veuronal growth regulator 1 (IgLON family member 1)	NECR1
Jenritin	NRN1
Neuritin_like protein	NRN1I
5' nucleotidase (5' NT) (EC 3.1.3.5) (Ecto 5' nucleotidase) (CD entiren CD79)	NT5F
Neurotrimin (hNT) (IgLON family member 9)	NTM
neurotinnin (invi) (igLOW family member 2)	IN I IVI

Protein	Symbol
Netrin-G1 (Laminet-1)	NTNG1
Netrin-G2 (Laminet-2)	NTNG2
Oligodendrocyte-myelin glycoprotein	OMG
Opioid-binding protein/cell adhesion molecule (OBCAM) (OPCML) (Opioid-binding	OPCML
cell adhesion molecule) (IgLON family member 1)	
Otoancorin	OTOA
Urokinase plasminogen activator surface receptor (U-PAR) (uPAR) (Monocyte acti-	PLAUR
vation antigen Mo3) (CD antigen CD87)	
Placenta-expressed transcript 1 protein	PLET1
Prion-like protein doppel (PrPLP) (Prion protein 2)	PRND
Major prion protein (PrP) (ASCR) (PrP27-30) (PrP33-35C) (CD antigen CD230)	PRNP
Testisin (EC 3.4.21) (Eosinophil serine protease 1) (ESP-1) (Serine protease 21)	PRSS21
Serine protease 41 (EC 3.4.21) (Testis serine protease 1) (TESSP-1)	PRSS41
Prostate stem cell antigen	PSCA
UL-16 binding protein 5 (Retinoic acid early transcript 1G protein)	RAET1G
UL16-binding protein 6 (Retinoic acid early transcript 1L protein)	RAET1L
Reversion-inducing cysteine-rich protein with Kazal motifs (hRECK) (Suppressor of	RECK
tumorigenicity 15 protein)	
Repulsive guidance molecule A (RGM domain family member A)	RGMA
RGM domain family member B (DRG11-responsive axonal guidance and outgrowth	RGMB
of neurite) (DRAGON)	
Reticulon-4 receptor (Nogo receptor) (NgR) (Nogo-66 receptor)	RTN4R
Reticulon-4 receptor-like 1 (Nogo receptor-like 2) (Nogo-66 receptor homolog 2)	RTN4RL1
(Nogo-66 receptor-related protein 3) (NgR3)	
Reticulon-4 receptor-like 2 (Nogo receptor-like 3) (Nogo-66 receptor homolog 1)	RTN4RL2
(Nogo-66 receptor-related protein 2) (NgR2)	
Semaphorin-7A (CDw108) (JMH blood group antigen) (John-Milton-Hargen human	SEMA7A
blood group Ag) (Semaphorin-K1) (Sema K1) (Semaphorin-L) (Sema L) (CD antigen	
CD108)	
Acid sphingomyelinase-like phosphodiesterase 3b (ASM-like phosphodiesterase 3b)	SMPDL3B
(EC 3.1.4)	
Sperm acrosome membrane-associated protein 4 (Sperm acrosomal membrane-	SPACA4
associated protein 14)	
Hyaluronidase PH-20 (Hyal-PH20) (EC 3.2.1.35) (Hyaluronoglucosaminidase PH-20)	SPAM1
(Sperm adhesion molecule 1) (Sperm surface protein PH-20)	
Shadow of prion protein (Protein shadoo)	SPRN
Teratocarcinoma-derived growth factor 1 (Cripto-1 growth factor) (CRGF) (Epider-	TDGF1
mal growth factor-like cripto protein CR1)	
Alpha-tectorin	TECTA
Beta-tectorin	TECTB
Testis-expressed protein 101 (Cell surface receptor NYD-SP8) (Scleroderma-	TEX101
associated autoantigen) (Spermatogenesis-related gene protein)	
Tissue factor pathway inhibitor (TFPI) (Extrinsic pathway inhibitor) (EPI)	TFPI
(Lipoprotein-associated coagulation inhibitor) (LACI)	
Thy-1 membrane glycoprotein (CDw90) (Thy-1 antigen) (CD antigen CD90)	THY1
Tumor necrosis factor receptor superfamily member 10C (Antagonist decov receptor	TNFRSF10C
for TRAIL/Apo-2L) (Decov TRAIL receptor without death domain) (Decov receptor	11011001100
1) (DcB1) (Lymphocyte inhibitor of TRAIL) (TNE-related apoptosis-inducing ligand	
recentor 3) (TRAIL recentor 3) (TRAIL R3) (TRAIL recentor without an intracel	
lular domain) (CD antigen CD263)	
Trehalase (EC 3.2.1.28) (Alpha alpha-trehalase) (Alpha alpha-trehalose glucohydro	TREH
lase)	11(1/11
III.16-binding protein 1 (ALCAN-beta) (NKG2D ligand 1) (N2DL-1) (NKC2DL1)	ULBP1
(Retinoic acid early transcript 11)	
III.16-binding protein 2 (ALCAN-alpha) (NKG2D ligand 2) (N2DL-2) (NKC2DL2)	ULBP2
(Retinoic acid early transcript 1H)	
UL16-binding protein 3 (ALCAN-gamma) (NKG2D ligand 3) (N2DL-3) (NKG2DL3)	ULBP3
(Retinoic acid early transcript 1N)	

Protein	Symbol	
Uromodulin (Tamm-Horsfall urinary glycoprotein) (THP) [Cleaved into: Uromodulin,	UMOD	
secreted form]		
Pantetheinase (EC 3.5.1.92) (Pantetheine hydrolase) (Tiff66) (Vascular non-	VNN1	
inflammatory molecule 1) (Vanin-1)		
Vascular non-inflammatory molecule 2 (Vanin-2) (EC 3.5.1.92) (Glycosylphosphatidyl	VNN2	
inositol-anchored protein GPI-80) (Protein FOAP-4)		
Vascular non-inflammatory molecule 3 (Vanin-3) (EC 3.5.1.92)	VNN3	
Xaa-Pro aminopeptidase 2 (EC 3.4.11.9) (Aminoacylproline aminopeptidase)	XPNPEP2	
(Membrane-bound aminopeptidase P) (Membrane-bound APP) (Membrane-bound		
AmP) (mAmP) (X-Pro aminopeptidase 2)		

Table $S3$:	Phenotypic	comparison	of	Synthesis+Transamidase
(S/T) vs. R	emodeling gr	roups		

HP:0000079 Abnormality of the urinary sys- $35/113(31.0\%) 1/39(2.6\%)$ 12.	.946 0.04648
tem	
HP:0010935 Abnormality of the upper urinary $34/113 (30.1\%) 0/39 (0.0\%)$ 15.	.116 0.01466
tract	
HP:0000708 Behavioral abnormality 15/113 (13.3%) 24/39 (61.5%) 35.	.407 0
HP:0012758 Neurodevelopmental delay 72/113 (63.7%) 39/39 (100.0%) 19.	.377 0.00156
HP:0000750 Delayed speech and language de- 34/113 (30.1%) 30/39 (76.9%) 26.	.089 0.00005
velopment	
HP:0001270 Motor delay $22/113 (19.5\%) 29/39 (74.4\%) 39.$.181 0
HP:0040195 Decreased head circumference 21/113 (18.6%) 19/39 (48.7%) 13.	.577 0.03319
$HP:0000252 Microcephaly \qquad \qquad 20/113 \ (17.7\%) 19/39 \ (48.7\%) \qquad 14.$.625 0.01902
$HP:0005484 Postnatal \ microcephaly \qquad 13/113 \ (11.5\%) 16/39 \ (41.0\%) \qquad 16.$.366 0.00757
HP:0000598 Abnormality of the ear $27/113 (23.9\%) 22/39 (56.4\%) 14.$.033 0.02604
HP:0031703 Abnormal ear morphology $21/113 (18.6\%) 21/39 (53.8\%) 18.$.029 0.00316
HP:0000356 Abnormality of the outer ear $18/113 (15.9\%) 21/39 (53.8\%) 21.$.853 0.00043
HP:0000377 Abnormality of the pinna $17/113 (15.0\%) 18/39 (46.2\%) 15.$.831 0.01004
HP:0100886 Abnormality of globe location $26/113 (23.0\%) 22/39 (56.4\%) 14.$.971 0.01583
$HP:0000316 Hypertelorism \qquad \qquad 23/113 \ (20.4\%) 20/39 \ (51.3\%) \qquad 13.$.671 0.03158
HP:0030669 Abnormal morphology of the oc- $20/113(17.7\%)$ $21/39(53.8\%)$ 19.	.232 0.00168
$ular \ adnexa$	
HP:0000492 Abnormal eyelid morphology $19/113 (16.8\%) 20/39 (51.3\%) 18.$.058 0.00311
HP:0008050 Abnormality of the palpebral fis- $13/113 (11.5\%) = 18/39 (46.2\%) = 21.$.44 0.00053
sures	
HP:0200007 Abnormal size of the palpebral 7/113 (6.2%) 14/39 (35.9%) 21.	.483 0.00052
fissures	
HP:0000422 Abnormality of the nasal bridge $23/113 (20.4\%) 22/39 (56.4\%) 18$.	.086 0.00306
HP:0000431 Wide nasal bridge $13/113 (11.5\%) 22/39 (56.4\%) 32.$.987 0
HP:0000159 Abnormality of the lip $32/113 (28.3\%) 24/39 (61.5\%) 13.$.751 0.03027
HP:0000177 Abnormality of upper lip $28/113 (24.8\%) 24/39 (61.5\%) 17.$.407 0.00437
HP:0011339 Abnormality of upper lip vermil- $23/113 (20.4\%) = 24/39 (61.5\%) = 23.$.023 0.00023
lion	
HP:0010804 Tented upper lip vermilion $20/113 (17.7\%) 21/39 (53.8\%) 19.$.232 0.00168
HP:0100737 Abnormality of the hard palate $6/113(5.3\%)$ $17/39(43.6\%)$ 33.	.084 0
HP:0000202 Oral cleft $6/113 (5.3\%) = 18/39 (46.2\%) = 36.$.377 0
HP:0000175 Cleft palate $6/113 (5.3\%) = 17/39 (43.6\%) = 33.$.084 0
HP:0001939 Abnormality of $54/113 (47.8\%) 32/39 (82.1\%) 13$.	.855 0.02863
metabolism/homeostasis	
HP:0012379 Abnormal enzyme/coenzyme ac- 37/113 (32.7%) 31/39 (79.5%) 25.	.624 0.00006
tivity	
HP:0004379 Abnormality of alkaline phos- 37/113 (32.7%) 31/39 (79.5%) 25.	.624 0.00006
phatase activity	
HP:0003155 Elevated alkaline phosphatase 32/113 (28.3%) 31/39 (79.5%) 31.	.28 0

Table **S4**: Publications curated for this work

Author	Title	Year	PMID
Abdel-Hamid	PGAP3-related hyperphosphatasia with mental re-	2017	28390064
	tardation syndrome: Report of 10 new patients and		
	a homozygous founder mutation.		
Almeida	Hypomorphic promoter mutation in PIGM causes	2006	16767100
.	glycosylphophatidylintosol deficiency		
Barone	DPM2-CDG: a muscular dystrophy-	2012	23109149
	dystroglycanopathy syndrome with severe epilepsy.	2014	
Belet	Early frameshift mutation in PIGA identified a large	2014	24357517
	XLID family without neonatal lethality.	0015	05004400
Bosch	Cerebral visual impairment and intellectual disabil-	2015	25804403
	ity caused by PGAP1 variants.	0014	04050109
Brady	Exome sequencing identifies a recessive PIGN splice	2014	24852103
	site mutation as a cause of syndromic congentital		
Comon: ID1	diaphragmatic nernia	2019	20472025
Ceroni JR1	Large deletion in PIGL: a common mutational mech-	2018	29473937
Chivonoby	ansni ni Onivili Syndronie: Clycosylphophatidylinosital (CDI) anchar deficiency	2014	94967057
Ullyonobu	caused by mutations in PICW is associated with	2014	24307037
	WEST syndrome and hyperphosphatasia with mon		
	tal retardation syndrome		
Edvarson	Mutations in the phosphatidulinosital glucan C	2017	27604521
Euvaison	(PICC) gong are associated with opilopsy and intel	2017	21094021
	lectual disability		
Fuiiwara	Mutations in PICL in a patient with Mahry syn-	2015	
rujiwara	drome	2010	
Granzow	Loss of function of PGAP1 as a cause of severe en-	2015	26050939
Granzow	cephalopathy identified by Whole Exome Sequenc-	2010	20000000
	ing: Lessons of the bioinformatics pipeline.		
Hansen	Hypomorphic mutations in PGAP2, encoding a	2013	23561846
	GPI-anchor-remodeling protein, cause autosomal-		
	recessive intellectual disability.		
Hogrebe	A novel mutation in PIGW causes glycosylphos-	2016	27626616
0	phatidylinositol deficiency without hyperphosphata-		
	sia		
Horn	Delineation of PIGV mutation spectrum and associ-	2014	24129430
	ated phenotypes in hyperphosphatasia with mental		
	retardation syndrome		
Howard	Mutations in PGAP3 impair GPI-anchor matura-	2014	24439110
	tion, causing a subtype of hyperphosphatasia with		
	mental retardation.		
Ilkovski	Mutations in PIGY: expanding the phenotype of in-	2015	26293662
	herited glycosylphophatidylinosol deficiencies		
Jezela-Stanek	Congenital disorder of glycosylphosphatidylinositol	2016	26879448
	(GPI)-anchor biosynthesis–The phenotype of two pa-		
	tients with novel mutations in the PIGN and PGAP2		
- 1	genes	0.045	
Johnston	The phenotype of a germline mutation in PIGA:	2012	22305531
	the gene somatically mutated paroxysmal nocturnal		
T 1	hemoglobinuria	2015	00000
Johnstone	Compound heterozygous mutations in the gene	2017	28334793
	PIGP are associated with early infantile epileptic en-		
TZ .	cephalopathy.	0014	0.180.001.0
Kato	PIGA mutations cause early onset epileptic en-	2014	24706016
	cephalopathies and distinctive features		

Table **S4** – Continued from previous page

Author	Title	Year	PMID
Kettwig	Compound heterozygous variants in PGAP1 caus-	2016	27206732
0	ing severe psychomotor retardation, brain atrophy,		
	recurrent apneas and delayed myelination: a case re-		
	port and literature review		
Kim	Dolichol phosphate mannose synthase (DPM1) mu-	2000	10642597
	tations define congenital disorder of glycosylation Ie		
	(CDG-Ie)		
Knaus	Rare Noncoding Mutations Extend the Mutational	2016	27120253
	Spectrum in the PGAP3 Subtype of Hyperphos-	-010	
	phatasia with Mental Retardation Syndrome.		
Krawitz	PGAP2 mutations, affecting the GPI-anchor-	2013	23561847
	synthesis pathway, cause hyperphosphatasia with		
	mental retardation syndrome		
Krawitz	Mutations in PIGO a member of the GPI-anchor-	2012	
111001102	synthesis pathway cause hyperphosphatasia with	_01_	
	mental retardation		
Kuki	B6-responsive epilepsy due to inherited GPI defi-	2013	24049131
I Yuki	ciency	2010	21010101
Kyamung	A novel intellectual disabilitysyndrome caused by	2013	23636107
irvainaing	GPI anchor deficiency due to homozygous mutations	2010	20000101
	in PIGT		
Lam	Expanding the clinical and molecular characteristics	2015	25943031
Lam	of PIGT-CDG a disorder of glycosylphosphatidyli-	2010	20040001
	nositol anchors		
Lefebeder	Deficiency of Dol-P-Man synthese subunit DPM3	2009	10576565
Leiebedei	bridges the congenital disorders of glycosylation with	2005	13370303
	the dystroglycanopathics		
Lin	We report the first family with PICA associated	2018	20656008
17111	apiloptic oncorbalopathy in Taiwan and hope to alu	2018	29000098
	cidate its special phonotype and inheritance pattern		
Makrythanacie	Pathogonic Variants in PICC Cause Intellectual Dis	2016	26006048
Maki y manasis	ability with Soiguros and Hypotonia	2010	20330340
Montin	Clinical whole—generate accurating in severe early	2014	
11/1/11/111	onact opilongy reveals new genes and improves males	2014	
	onset epilepsy reveals new genes and improves molec-		
M J	ular diagnosis	0011	01402057
Maydan	Multiple congenital anomalies-hypotomia-seizures	2011	21493937
мт т	syndrome is caused by a mutation in PIGN	0010	07090415
McInerney-Leo	tions in DICN in two Separate Equilies	2010	27038415
۸ <i>۲</i> :	Null neutotion in DCAD1 investige California en el constat	9014	04704195
Murakami	Null mutation in PGAP1 impairing Gpi-anchor mat-	2014	24784135
	uration in patients with intellectual disability and		
Nalsonauma	DICO mutations in intractable children and gaven	2014	94417746
пакашига	developmental dalars with wild about in a faller line	2014	24417740
	developmental delay with mild elevation of alkaline		
NT - 1 1	Normal account of the tensor DICT montations	9014	94006049
Nakashima	Novel coumputed neterozygous PIG1 mutations	2014	24900948
	caused multiple congenital anomalies-hypotonia		
NT (1··	seizures syndrome 3	0017	00704014
Nampoothiri	Hyperphosphatasia with Mental Retardation Syn-	2017	28794914
N	drome Due to a Novel Mutation in PGAP3	0010	05051400
Naseer	A novel mutation in PGAP2 gene causes develop-	2016	27871432
	mental delay, intellectual disability, epilepsy and mi-		
	crocephaly in consanguineous Saudi family.	0010	
Ng	Mutations in the glycosylphosphatidylinositol gene	2012	
	PIGL cause CHIME syndrome.		
Nguyen	Mutations in GPAA1, Encoding a GPI Transami-	2017	29100095
	dase Complex Protein, Cause Developmental Delay,		
	Epilepsy, Cerebellar Atrophy, and Osteopenia.		

Table **S4** – Continued from previous page

Author	Title	Year	PMII
Nguyen	A PIGH mutation leading to GPI deficiency is asso-	2018	29603516
	ciated with developmental delay and autism.		
Ohba	PIGN mutations cause congenital anomalies, devel-	2014	24253414
	opmental delay, hypotonia, epilepsy, and progressive		
D	cerebellar atrophy		
Pagnamenta	A homozygous variant disrupting the PIGH start-	2018	PMID: 29573052
	codon is associated with developmental delay,		
Demomente	epilepsy, and microcephaly.	2017	0020757
ragnamenta	ancher biogenesis defects are a rare cause of devel	2017	2032131
	opmental disorders		
Perez	A Bare Variant in PGAP2 Causes Autosomal Re-	2017	2911910
1 01 02	cessive Hyperphosphatasia with Mental Retardation	-011	_011010
	Syndrome, with a Mild Phenotype in Heterozygous		
	Carriers.		
Sakaguchi	A novel PGAP3 mutation in a Croatian boy with	2017	2953177
	brachytelephalangy and a thin corpus callosum.		
Schenk	MPDU1 mutations underlie a novel human congeni-	2001	1173356
	tal disorder of glycosylation, designated type If.		
Swoboda	A novel germline PIGA mutation in Ferro-Cerebro-	2014	2425928
	Cutaneous syndrome: a neurodegenerative X-linked		
	epileptic encephalopathy with systemic iron-over-		
Tamila Creare	10ad The geneturie and pheneturie spectrum of DICA de	2015	
Tarano-Graovac	ficionev	2015	
Thiffault	Hypotonia and intellectual disability without dys-	2017	2909660
1 miliadit	morphic features in a patient with PIGN-related dis-	2011	2000000
	ease		
Thompson	Phenotypic variability in hyperphosphatasia with	2012	2231519
-	seizures and neurologic deficit (Mabry syndrome).		
Van den Bergh	A homozygous DPM3 mutation in a patient with	2017	2880381
	alpha-dystroglycan-related limb girdle muscular dys-		
	trophy		
Van der Crabben	Expanding the spectrum of phenotypes associated	2014	2425918
	with germline PIGA mutations: a child with devel-		
	opmental delay, accelerated linear growth, facial dys-		
	morphisms, elevated alkaline phosphatase, and pro-		
Williams	Additional evidence that PCAP1 loss of function	2015	2582341
vv mams	causes autosomal recessive global developmental de-	2010	2002041
	lay and encephalopathy.		
Xie LL	A novel germline PIGA mutation causes early-onset	2018	2950286
	epileptic encephalopathies in Chinese monozygotic		
	twins.		
Yang	Congenital disorder of glycosylation due to DPM1	2013	2385642
	mutations presenting with dystroglycanopathy-type		
	congenital muscular dystrophy.		
Zehavi	A homozygous PIGO mutation associated with se-	2017	2890081
	vere infantile epileptic encephalopathy and cor-		
	pus callosum hypoplasia, but normal alkaline phos-		
	phatase levels.		