**Supplemental Data**

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| --- | --- | --- | --- | --- |
| **Gene** | **Phenotype MIM number** | **Phenotype (based on OMIM classification)** | **Inheritance** | **First report** |
| *GPAA1* | #617810 | Glycosylphosphatidylinositol biosynthesis defect 15 | AR | 1 |
| *PIGA* | #300868 | Multiple congenital anomalies-hypotonia-seizures syndrome 2 (somatic mutations associated with Paroxysmal nocturnal hemoglobinuria 1; #300818) | XLR | 2 |
| *PGAP1* | #611655 | Mental retardation, autosomal recessive 42 | AR | 3 |
| *PGAP2* | #614207 | Hyperphosphatasia with mental retardation syndrome 4 | AR | 4 |
| *PGAP3* | #615716 | Hyperphosphatasia with mental retardation syndrome 3 | AR | 5 |
| *PIGC* | #601730 | Glycosylphosphatidylinositol biosynthesis defect 16 | AR | 6 |
| *PIGG* | #616917 | Mental retardation, autosomal recessive 53 | AR | 7 |
| *PIGH* | #618010 | Glycosylphosphatidylinositol biosynthesis defect 17 | AR | 8 |
| *PIGL* | #280000 | CHIME syndrome | AR | 9 |
| *PIGM* | #610293 | Glycosylphosphatidylinositol biosynthesis defect 1 | AR | 10 |
| *PIGN* | #614080 | Multiple congenital anomalies, hypotonia, seizures syndrome 1 | AR | 11 |
| *PIGO* | #614749 | Hyperphosphatasia with mental retardation syndrome 2 | AR | 12 |
| *PIGP* | #617599 | Epileptic encephalopathy, early infantile, 55 | AR | 13 |
| *PIGQ* | Not Available | Epileptic encephalopathy, early infantile | AR | 14 |
| *PIGS* | #618143 | Glycosylphosphatidylinositol biosynthesis defect 18 | AR | 15 |
| *PIGT* | #615398 | Multiple congenital anomalies‐hypotonia‐seizures syndrome 3 | AR | 16 |
| *PIGV* | #239300 | Hyperphosphatasia with mental retardation syndrome 1 | AR | 17 |
| *PIGW* | #616025 | Hyperphosphatasia with mental retardation syndrome 5 | AR | 18 |
| *PIGY* | #616809 | Hyperphosphatasia with mental retardation syndrome 6 | AR | 19 |

**Supplemental Table e-1:** Genes associated with Inherited GPI Deficiencies by germline variants.

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
|  | **Median fluorescence intensity (MFI)** | | | | |
| **Sample ID** | **CD55** | **CD59** | **CD16** | **FLAER** | **CD24** |
| Patient 2 | 6640 | 5384 | **54348\*** | 29920 | 3003 |
| Patient 4 | 5317 | 7358 | **40142\*\*** | 26788 | 1822 |
| Control subject 1 | 7964 | 5732 | 94848 | 21530 | 4937 |
| Control subject 2 | 5674 | 6247 | 85267 | 30612 | 5547 |
| Control subject 3 | 4861 | 4998 | 112165 | 23018 | 3157 |
| Control subject 4 | 6535 | 6264 | 107122 | 30198 | 5348 |
| Control subject 5 | 6246 | 4740 | 98171 | 17189 | 1999 |
| Control subject 6 | 3984 | 5366 | 97301 | 19927 | 2802 |
| Control subject 7 | 4024 | 4269 | 99099 | 20169 | 1841 |
| Control subject 8 | 5823 | 5547 | 84612 | 17220 | 2162 |
| Control subject 9 | 8280 | 4774 | 67231 | 16553 | 2536 |
| Control subject 10 | 4940 | 4129 | 69869 | 15251 | 2745 |
| Average MFI of controls (±SD) | 5833.1 ± 1477.6 | 5206.6 ± 753.3 | 91568.5 ± 14762.2 | 21166.7 ± 5421.7 | 3307.4 ± 1422.0 |

**Supplemental Table e-2:** Median fluorescence intensities (MFIs) of the 5 markers investigated by flow cytometry in circulating granulocytes of Patients 2 and 4, and in control subjects. Values with statistical significance are shown in bold; \*p=0.05; \*\*p=0.01. SD, standard deviation.

**Supplemental References:**

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