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Supplemental Data

Hypomorphic Mutations in *PGAP2*, Encoding a GPI-Anchor-Remodeling Protein, Cause Autosomal-Recessive Intellectual Disability

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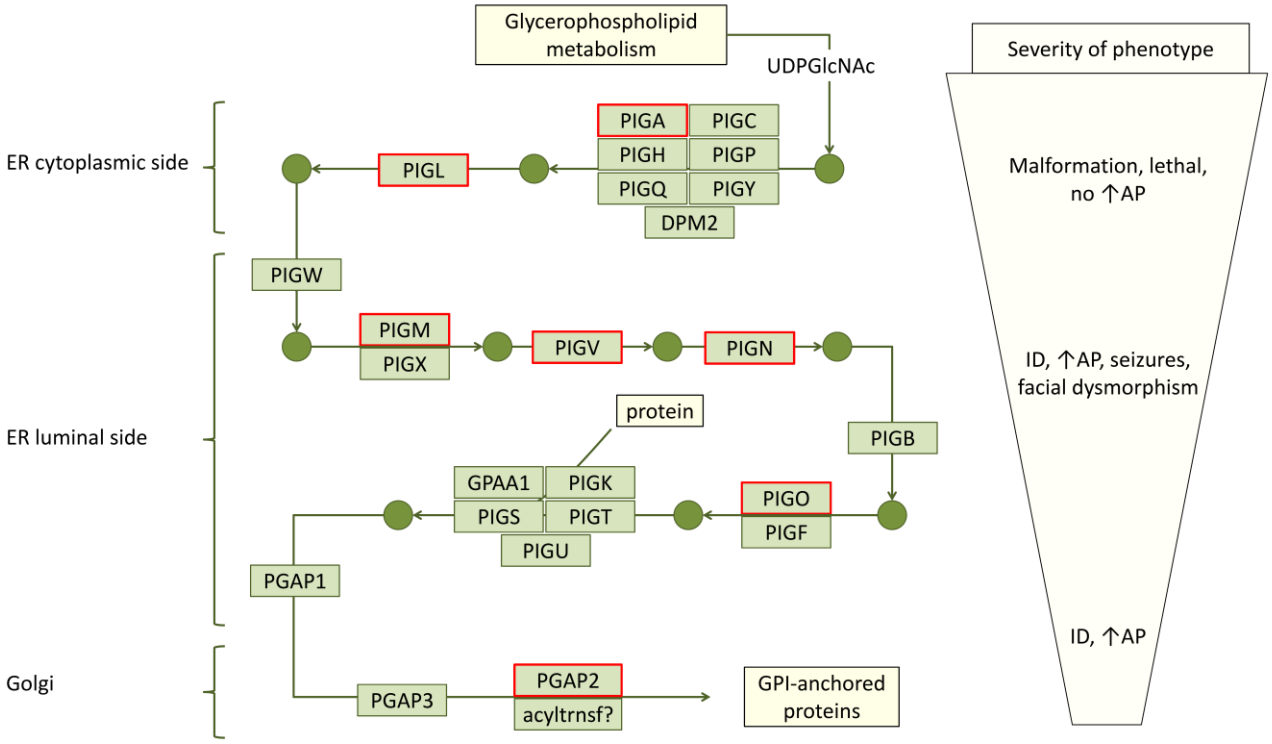

Figure S1. Simplified GPI-Anchor Biosynthesis Pathway Based on KEGG[27] and Kinoshita et al., 2008[1]

Genes with reported mutations are in red boxes, more details on phenotypes and the mutations are in Table 1. There is a tendency of milder phenotype toward the downstream end of the pathway (see discussion). ID: intellectual disability, AP: alkaline phosphatase, ER: endoplasmic reticulum

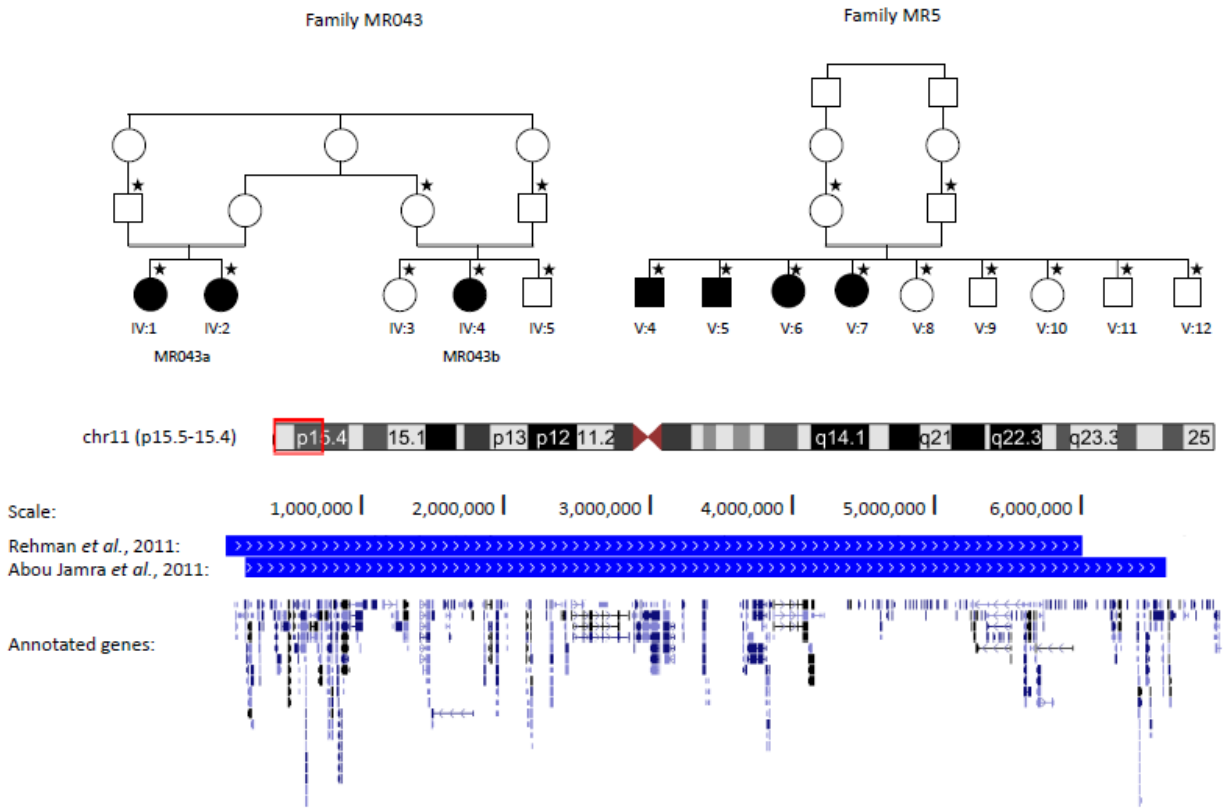

Figure S2. Pedigrees of Examined Families and Linkage Findings

Top: The pedigrees of the Pakistani MR5 and the Syrian MR043 families persons whose DNA was available are marked with asterisks.

Bottom: The blue bars represent the overlapping linkage regions with significant LOD scores,[12; 13] the homozygosity region with genes was modified from UCSC genome browser.

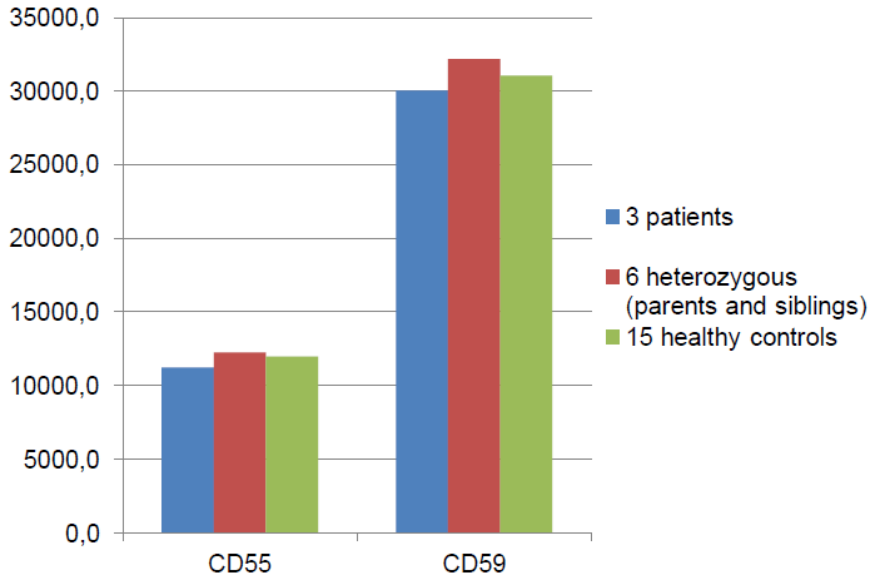

Figure S3. Results of FACS with LCLs of Affected Persons from Family MR043, Heterozygous Family Members, and Healthy Controls

The differences of the mean fluorescence intensity (MFI, Y axis) between the homozygous, heterozygous, and control persons were not significant.