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The clinical phenotype of PIGN deficiency and consequences of defective GPI biogenesis

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The PIGN gene (MIM606097) encodes glycosylphosphatidylinositol (GPI) ethanolamine phosphate transferase-1 which adds the ethanolamine phosphate to the first mannose on the GPI anchor. GPI anchoring of proteins is a highly conserved process present in most eukaryotic cells. GPI-anchored proteins perform a diverse set of functions including roles in signal transduction, cell adhesion and antigen presentation. To date mutations in 12 genes involved in GPI anchor synthesis have been associated with human disease. Maydan first described mutations in this gene in a consanguineous Israeli family of Palestinian origin; children presented with multiple congenital anomalies, severe neurological involvement and early death. Four additional cases have been reported since then. We report three unrelated patients with mutations in PIGN and diverse clinical characteristics. Patients 1 and 2 presented during their newborn period with hypotonia, intractable epilepsy, facial dysmorphism as well as gastrointestinal, genitourinary and skeletal anomalies including brachytelephalangy. Patient 1 had metopic suture craniosynostosis and left choanal atresia. Patient 2 had an imperforate anus, uterine didelphys and a small splenic cyst. Patient 3 with a milder phenotype manifested as hypotonia, developmental delay, dysmorphic facial features and early onset refractory epilepsy. Fluorescence activated cell sorting analysis was performed on cultured skin fibroblast lines from Patients 2 and 3 which demonstrated decreased cell surface expression of the GPI anchored protein CD59, consistent with deficient GPI anchor biogenesis in these cells. Mechanistic studies of PIGN deficiency are currently underway in our laboratory.

Biography

Francis Jeshira Reynoso has completed her Pediatrics Residency at Lincoln Medical Center; Bronx NY affiliated to Weill Cornell Medical College. She is currently a Clinical Genetics Fellow at the Children's Hospital of Philadelphia and is conducting research under the mentorship of Miao He and her PhD is focused on Congenital Disorders of Glycosylation.

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