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Assessment and Early Intervention of Speech and Language in a Child with Congenital Disorder of Glycosylation

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Congenital disorders of glycosylation (CDG), or carbohydrate deficient glycoprotein syndrome forms a cluster of multisystem disorders characterised by defective glycoprotein biosynthesis, due to various biochemical mechanisms. This is often manifested as a syndromic condition. Phosphomannomutase2 (PMM2) CDG is the most common form of CDG, also known as CDG type 1a.Symptoms associated with PMM2 CDG are broad and highly variable. Prominent symptoms include psychomotor retardation, underdevelopment (hypoplasia) of the cerebellum, crossed eyes (strabismus) and facial dysmorphism. Other secondary symptoms are characterised by feeding problems, neuromuscular abnormalities, severe developmental delay with varying degree of cognitive impairment, and symptoms resulting from episodes of internal organ failure, or additional physical abnormalities. Development of early communication abilities is compromised due to the above clinical manifestations.

This single-subject case study will describe the clinical presentation of a 1-year old male child who had an established diagnosis of CDG type 1a based on genetic evaluation. The child presented with delayed development of speech and language milestones, delayed vision maturation and reduced hearing sensitivity in both ears. This case study will focus on the challenges of assessing speech, language and hearing in such scenario. The importance of team approach in establishing the baseline to implement early intervention in such scenario will be highlighted.