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Novel causative mutation in the gene for Galloway-Mowat syndrome has been identified. Osgep (c.2S G>A p.GLySer) has not been reported in the international database - Report of case and literature review

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Introduction: Galloway- Mowat syndrome is a rare hereditary renal, neurological disease characterized by microcephaly, intellectual disability, hiatus hernia, skeletal anomalies, and nephrotic syndrome. It appears to be transmitted as an autosomal recessive trait. Recently, novel causative mutations for this disease have been identified in the gene-encoding subunit OSGEP. The gene variant has not been reported before in the international database.

Case Presentation: A twenty months old Egyptian with working diagnosis of Galloway- Mowat syndrome caused by OSGEP gene (c.25 G>A p.GlySer). She was born at term by caesarean section due to twin delivery. Birth weight was 2700g. She was born with normal head circumference and weight. At the age of 3 months, mother noticed that her head circumference is not increasing compared to her twin, her current HC <3rd centile. This girl displayed various features of facial dysmorphism (microcephaly, deeply sited eyes, and high arched palate). In addition, she has spasticity, hyperreflexia, truncal hypotonia, Global developmental delay, failure to thrive and epileptic disorders. Renal ultrasound revealed bilateral early to grade 1 renal parenchymatous pathological changes. Her serum creatinine levels were 17 umol/L (low). The segregation analysis showed that both parents and her twin are carriers which supports that the variant of OSGEP is likely to be pathogenic.

Methods: This study was designed as a case report using patient clinical manifestation with a literature review, together with family study through segregation analysis that can yield robust data to re-classify a variant of unknown clinical significance.

Results: The OSGEP gene (c.25 G>A p.GlySer) is most likely pathogenic from the patient phenotype and family segregation data. However, gene functioning is the gold standard method to classify this variant which is still under process.

Conclusions: We report a familial Galloway-Mowat syndrome caused by the OSGEP gene (c.25 G>A p.GlySer) with both parents and her twin carrying a novel heterozygous. She displayed various features; microcephaly, deeply sited eyes, high arched palate, spasticity, hyperreflexia, truncal hypotonia, Global developmental delay, failure to thrive and epileptic disorders.

Biography

Aisha Khameis Ahmed is a third-year pediatric resident in the department of pediatrics at Fujairah Hospital, UAE. She completed MBBS from the UAE University. Before joining the pediatric residency program, she worked as a General practitioner for over seven years in pediatrics.

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