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The clinical manifestation and genetic implication of Baraitser-Winter syndrome Type 2: A case report

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Introduction: Baraitser-Winter Syndrome Type 2 is a rare condition with fewer than 8 cases reported in medical literature. The major clinical anomalies are striking dysmorphic facial features with hypertelorism, broad nose with a large tip and prominent root, congenital non-myopathic ptosis, ridged metopic suture and arches eyebrows. Iris or retinal coloboma is present in many cases, in additional to sensorineural deafness and microcephaly. Early muscular involvement may develop over time, occasionally accompanied by congenital arthrogryposis. Literature has reported that, with the exception of one case, all reported patients with Baraitser-Winter Syndrome Type 2 have been sporadic. Whole-exome sequencing has been performed and identified heterozygous mis-sense changes in the cytoplasmic actin-coding gene *ACTG1*, a majority in which occurred, *de novo*. We report a young male with clinical features that are characteristic for Baraitser-Winter Syndrome-2whole exome genome sequencing that discovered mutations in the ACTB1 gene.

Case Study: An 18-year old Caucasian male was born by caesarian section and weighed 5 lb 14 oz at birth after a normal pregnancy in which there were no perinatal problems. His mother initially became concerned when she noticed he was experiencing some developmental delays and a mild floppy tone around 4 to 5 months, in addition to facial features that resembled some form of mental retardation. This patient's older brother also experienced similar developmental symptoms and facial features that presented around the same age-period as our patient. It was initially thought to be a form of Down's syndrome; however, both the patient and his brother tested negative for Down's syndrome on chromosomal analyses. Doctors had begun to think the patient had some form of autism spectrum disorder, but were unable to specifically confirm this. Now at the age of 18 years old, the patient has no understandable speech with distinctive facial features such as a broad nasal bridge and prominent epicanthic folds, lissencephaly, smaller than average head size, intellectual disability and hearing loss.

Discussion: Baraitser-Winter syndrome 2, a unique variant that is clinically similar to Baraitser-Winter syndrome, has only 8 cases that have been reported. Fewer than 50 cases of children with the characteristic clinical features resembling that of Baraitser-Winter Syndrome have been reported in the medical literature. Consequently, the phenotypic spectrum has broadened and includes microcornea, microphthalmia, arched eyebrows, gyral malformation, seizures, hypotonia, sensorineural deafness, congenital arthrogryposis and progressive joint stiffness. Cleft lip and palate, hallux duplex, congenital heart defects and renal tract anomalies are seen in some cases. The genetic mechanism behind the autosomal dominant Baraitser-Winter Syndrome has recently been associated with gain-of-function *de novo* mutations in the actin-encoding genes ACTB (Baraitser-Winter syndrome) and ACTG1 (Baraitser-Winter syndrome-2) that encode B- and Y-actins. These actin proteins are among the most important proteins in the function of individual cells and are expressed in cells throughout the body and are critical for cell division, cell movement and signaling.

Biography

Tanya C Allawh has completed her BS from the Pennsylvania State University and is completing her Post-doctoral studies at the Edward Via College of Osteopathic Medicine.

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