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Beckwith-Wiedemann Syndrome: Clinical Manifestations in a Preterm Infant Born at 33 Weeks' Gestation

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Morphological appearance of the macrosomic premature newborn at birth.

Morphological appearance of the infant at 2 months of age, weighing 5020g.

Figure

An infant aged 2 months, formerly macrosomic, was born prematurely at 33 weeks of gestation. The patient is female and was delivered vaginally in a monitored twin pregnancy, with an Apgar score of 10/10 and a birth weight of 2575g.

She was hospitalized during the neonatal period for respiratory distress and early bacterial neonatal infection due to prematurity at 33 weeks, receiving antibiotic coverage for 10 days. Physical examination revealed funnel chest (pectus excavatum), and musculoskeletal examination indicated long fingers and toes. Abdominal ultrasound showed bilateral ureterohydronephrosis, with normal echocardiograms (ETT and ETF). The ultrasound findings indicated dilation of the bilateral pyelocaliceal cavities without visible obstruction, suggestive of a junction syndrome.

At 1 month of age, the mother noted excessive growth along with macroglossia. Clinical examination revealed a hypotonic infant weighing 5020g (+two standard deviations) and measuring 61 cm (+two SD), exhibiting macroglossia and abdominal distension with an umbilical hernia. Ultrasound of the hips revealed congenital hip dislocation.

Beckwith-Wiedemann syndrome is suspected based on clinical signs such as macroglossia, macrosomia, and umbilical hernia, along with paraclinical findings like pyelocaliceal dilation. Genetic testing confirmed the diagnosis by detecting an anomaly on the long arm of chromosome 11, affecting the CDKN1C gene, which corroborates our diagnosis.

Beckwith-Wiedemann syndrome (BWS) is a rare genetic disorder named after the doctors who first described it, Dr. John Bruce Beckwith and Dr. Hans-Rudolf Wiedemann, in the early 20th century. This syndrome is characterized by a variety of clinical manifestations, including rapid growth leading to abnormal height for age, macroglossia (enlargement of the tongue), a predisposition to embryonal tumors, body asymmetries, and other congenital anomalies. Children with BWS exhibit genetic features such as genomic imprinting alterations, along with chromosomal duplications or microdeletions.

Management of BWS often involves a multidisciplinary approach, including medical care, surgical intervention, and health monitoring. Treatment may include surgery to correct congenital anomalies and regular surveillance to detect potential complications [1,2].

Bibliography

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