

## **Prenatal Diagnosis and Postnatal Outcome of 7q11.23 Duplication in a Fetus with Renal Pelvic Dilatation**

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7q11.23 duplication syndrome is a multisystemic developmental disorder characterized by variable manifestations, such as speech delay, mild craniofacial anomalies with distinctive facial features, and intellectual ability ranging from mental retardation to normal cognitive development. Approximately 30% of individuals with 7q11.23 duplication have one or more congenital anomalies. Penetrance is complete with variable expression of phenotypic features. Prevalence has been estimated at 1:7,500-1:20,000. The 7q11.23 duplication is frequently inherited from a parent.

Here we report a 33-year-old woman referred for prenatal diagnosis at 29 weeks of gestation due to fetal renal pelvic dilatation.

Chromosomal microarray analysis (CMA) was performed after a normal molecular rapid aneuploidy test result and identified a 1.44 Mb duplication at 7q11.23 - arr [GRCh37] 7q11.23 (72,700,467-74,136,633)x3 - overlapping the 7q11.23 duplication syndrome region, in a female fetus. The gain was inherited from the mother which had no previous clinical evaluation but a later reassessment revealed mild cognitive delay and language impairment.

Delivery was at 35 weeks due to a maternal respiratory infection with acute pulmonary edema. Newborn resuscitation was required for neonatal respiratory depression with an Apgar score of 1'-2, 5'-4, 10 '-8. Birth weight and length was 2140g and 43cm respectively, with a head circumference of 32cm.

In the neonatal period a transient systolic murmur was identified with no alterations on the echocardiogram. Renal and bladder ultrasound showed pelvic dilatation with no changes of the ureteral tract, suggesting a relation with ureteropelvic junction syndrome. Left pyeloplasty for the ureteropelvic junction syndrome was performed at 14 months of age.

Clinical evaluation at the age of 22 months revealed psychomotor development delay with delayed speech, facial features overlapping Williams-Beuren syndrome, and the systolic murmur grade I/VI was still present. Growth and weight were both normal.

To the best of our knowledge this is the second prenatal case of 7q11.23 duplication described. Although genitourinary tract abnormalities are not the most common feature in patients with 7q11.23 duplication, congenital anomalies of the urinary tract can occur in 15%-18%, including hydronephrosis and unilateral renal agenesis. This shows that the ultrasound abnormalities not always suggest a specific syndrome but after the identification of a pathogenic CNV made possible by the use of CMA a correlation may be achievable.

Additionally the discovery of CNVs in prenatal CMA may go beyond the context of the current pregnancy allowing for the identification of carriers thus having a larger impact in a family's health management and genetic counseling.