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Williams-beuren syndrome: 7q11.23 deletion

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Williams syndrome [OMIM 194050] (WS) also referred to as Williams-Beuren syndrome (WBS) is a multisystem genetic disorder and is characterized by typical facial dysmorphisms, congenital heart defects, mental retardation and a characteristic cognitive profile.WS occurs as a result of the de novo deletion of ≈1.55 to 1.83 Mb on chromosome 7q11.23. Familial cases can occur but are far less common than de novo cases. The deletion involves 26 to 28 genes, including the ELN gene, which codes for the protein elastin. Hemizygosity of the ELN gene coding for elastin has been demonstrated to be responsible for the vascular pathology in WS- arterial stenosis.

We present, a 3-year-old girl who is born to non consanguineous married couple with maternal age 26 years and paternal age 31 years. There was no significant medical and family history of note. The infant was born full term normal delivery. The birth weight was 2.5 kg with no perinatal issues. There was history of mild motor delay. She was also noted to have neuro developmental problems such as neonatal seizures and infantile spasms, developmental delay, speech and language delay and attention deficit hyperactivity disorder. On clinical examination the girl was overfriendly active child with facial dysmorphism. This included broad forehead, short upturned nose and protruding lips and a pointed chin. The proband also had failure to thrive and temperamental abnormality. Echocardiogram was done which revealed a large ventricular septal defect (VSD), bilateral peripheral pulmonary artery stenosis, mild supra valvular aortic stenosis (SVAS). Genetic analysis was performed employing FISH on metaphase chromosomes using LS1 WS Elastin gene region probe localized to 7q11.23 (VysisInc, USA). The result of FISH showed the red signal only on one of the chromosome 7 at 7q11.23 region in all the metaphases analyses suggesting deletion of ELN gene. Thereafter, the probandunderwent reparative surgery for her cardiovascular abnormalities: Doty"s procedure for SVAS, bilateral pulmonary artery patch plasty and fenestrated patch closure of VSD.

On follow up noted to have developed residual peripheral pulmonary artery stenosis and moderate aortic regurgitation. She received anticonvulsants, along with speech therapy, occupational therapy, play therapy and vocational training at child development centre as a part of management strategy for her neuro-developmental concerns.