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LETTER TO EDITOR

Unusual Brain Changes in Tetrasomy X Chromosomal Anomaly

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تغيرات غير عادية في الدماغ في حالة شذوذ الرباع الصبغي X

مشيلبوالا ، فينوجوبالان

Sir,

An 8-year-old girl presented with defective motor coordination, a speech delay, behavioural problems, and generalised hypotonia. On examination, she was found to have hypermobile joints; a cardiac anomaly (patent ductus arteriosus); facial dysmorphism, and irregularly spaced, overcrowded teeth. Magnetic resonance imaging (MRI) of the brain performed as part of the neurologic work-up showed generalised hypodense white matter with multiple widely distributed foci of high signal intensity on the T2 and flair sequences [ ]. On T1 sequences, these areas were hypointense. Subsequent investigations confirmed an underlying rare chromosomal anomaly called tetrasomy X (48, XXXX).

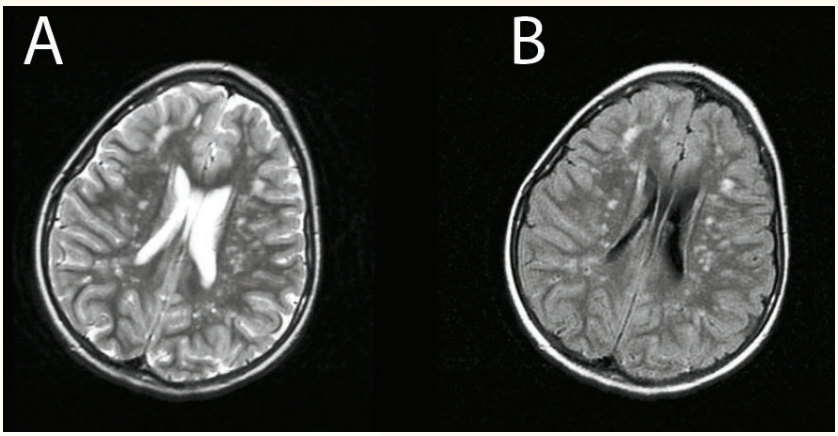

Figure 1: Magnetic resonance imaging of the brain in a child with tetrasomy X showing diffuse ill-defined areas of high density on a T2 weighted image (**A**) and an axial flare image (**B**). The corresponding areas were more lucent on the T1 weighted images.

Tetrasomy X is a rare chromosomal anomaly that was first reported in 1961 in two children with neuro-disabilities.[1–3] Although early case reports suggest mental deficiency as an obligatory feature of the syndrome, more recent reports confirm considerable variability in the phenotypical expression, making it difficult to assess the exact prevalence of the disorder.[4] In this 8-year-old girl, the presence of facial dysmorphism, a cardiac anomaly, and central hypotonia prompted the magnetic resonance imaging (MRI) brain scan as well as subsequent investigations that uncovered an otherwise unsuspected underlying disorder.

MRI changes associated with sex chromosome aneuploidies were extensively reviewed in a recent Australian publication.[5] Reported studies mostly involve children with Klinefelter's syndrome (47, XXY), and 47, XXY males. Findings in 47, XXY males include decreased grey and white matter volumes, with most pronounced effects in the frontal and temporal lobes. Functional studies have shown evidence of decreased lateralisation. It is widely believed that the severity of brain changes increases with the number of X chromosomes, as shown in children with Klinefelter's syndrome and varying numbers of X chromosomes. A report on 3 children with 49, XXXXY documented varying degrees of brain volume loss and white matter changes, and similar changes could be expected with 48, XXXX.[2] However, our patient exhibited widespread focal increase in the cerebral white matter density, with a normal cerebellum and brain stem. Although the exact relationship between the brain changes and neurobehavioural manifestations has not been elucidated, these changes are consistent with developmental aberrations, and could form the basis for the above manifestations in affected children.[5]

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