LETTER TO THE EDITOR

RING CHROMOSOME 8 WITH MOSAIC TRISOMY 8 SYNDROME IN AN INFANT

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Supernumerary ring chromosomes (SRCs) constitute a small category of the supernumerary marker chromosomes (SMCs), among which ring chromosome 8 is relatively rare (2). We report herein on an infant with mosaic trisomy of ring chromosome 8 in whom clinical features consisted of a mild form of mosaic trisomy 8 syndrome.

The patient was born by natural delivery at 40 weeks and 6 days, with a weight of 3,730 g. Postnatal suckling was languid, and activity was low. From about 10 days old, mild respiratory distress was seen during suckling. From 2 months old, repeated incidents of wheezing were noted. In the third month, the patient experienced repeated apneic attacks and was admitted to our hospital. During hospitalization, 5 or 6 apneic episodes lasting less than 10 sec/day were seen, but all resolved spontaneously. Electroencephalography showed normal results. Agenesis of the corpus callosum was seen on brain computed tomography (CT) (Fig. 1). Facial features included thick lips and a broad, upturned nose, and the patient had narrow shoulders and a slender trunk. Wrinkles in the limbs were deep. No heart or kidney malformations were evident. Chromosome analysis using peripheral lymphocytes was performed due to minor external malformation, and upon analysis of 30 cells by fluorescence *in situ* hybridization (FISH) with a chromosome 8 centromeric probe, mosaic 8 trisomy syndrome was diagnosed with mos47, XY, +r(8)[7]/46, XY[23].ish r(8) (D8Z2+) and ring chromosome 8 (Fig. 2). Chromosome analysis was not performed for the parents. Subsequently, the patient's breathing spontaneously stabilized. With regard to movement, he was able to sit at 8 months old, and could walk while holding on to something at 1 year and 2 months old. He could speak several meaningful words at 1 year and 2 months. His development is currently being observed.

Trisomy 8 mosaicism syndrome is a known entity, with clinical features of mild to severe mental and growth deficiency, facial dysmorphism, anomalies of the limbs such as palmar and plantar creases, camptodactyly and abnormal nails. Other abnormalities include urethral-renal,

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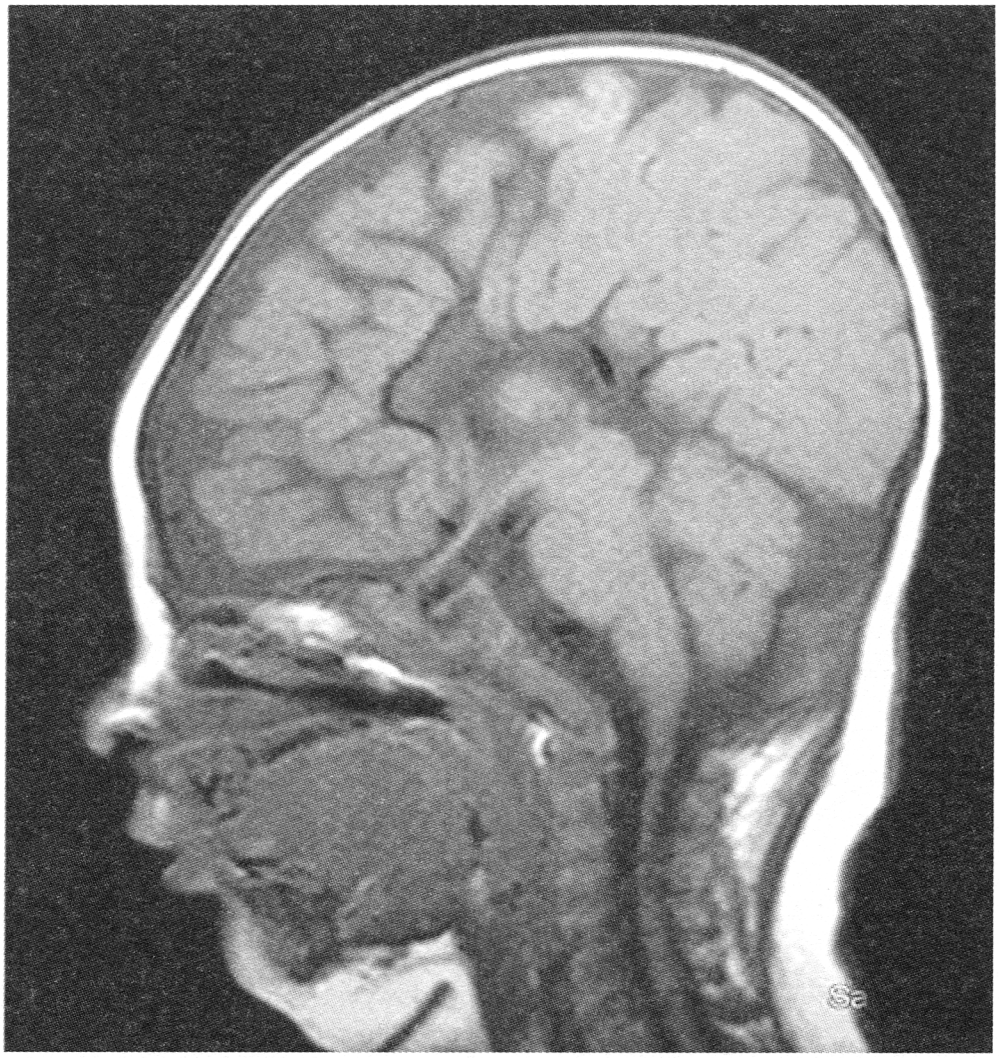

Figure 1: Suggital brain CT of the patient showed agenesis of corpus callosum.

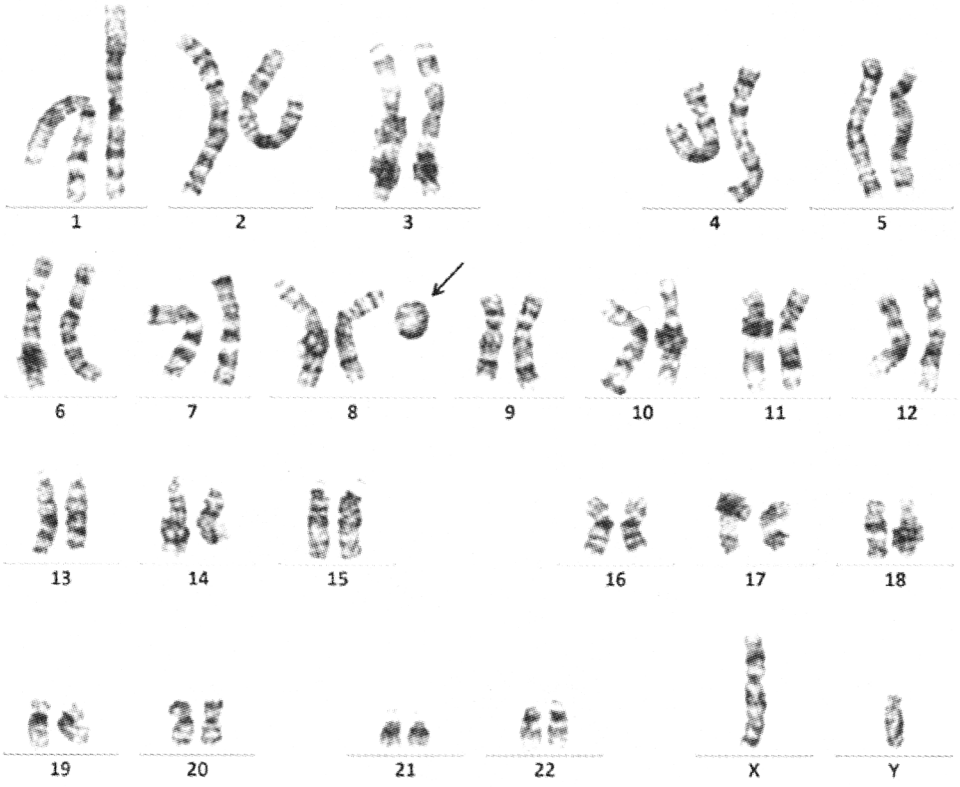

Figure 2: Chromosomal analysis employing with FISH methods of the patient revealed as follows; mos47,XY,+r(8)[7]/ 46,XY[23].ish r(8) (D8Z2+).

cardiac, skeletal defects, and brain anomalies (1). Based on the mosaic or non-mosaic condition, phenotype ranges from normality to various congenital anomalies, mostly overlapping with mosaic trisomy 8 syndrome (3).

The patient in this case was referred to the pediatric department at 3 months of age due to mild respiratory failure and poor suckling. His subsequent development was good, and at 14 months of age, he knew several meaningful words and could walk while holding on to something. Chromosome analysis using peripheral lymphocytes showed 47,XY, +r(8) in 7 of 30 cells, and the mosaic rate was 23.3%. Clinical manifestations of mosaic trisomy 8 syndrome have been reported by Eliana *et al.* for 26 patients (3) and Herry *et al.* for 19 patients (4). With regard to the genotype-phenotype correlation, both of these reports discussed the relationship with 8p or 8q deletion regions in a ring chromosome, but much remains unclear. Both of these reports also stated that, in relation to the mosaic rate, patients with a low mosaic rate in peripheral lymphocytes may tend to show clinically milder conditions. An earlier report by Rothenmund *et al.* (5) reported a 30-year-old man with normal phenotype in whom ring chromosome 8 was seen in 10% of peripheral lymphocytes. Herry *et al.* (4) reported an apparently normal 31-year-old woman with mosaic trisomy 8 in whom ring chromosome 8 was seen in 27% of peripheral lymphocytes. Our patient also has a low mosaic rate of 23% and is still 1 year old, but as of the time of writing, shows no delay in either movement or language; however, his future development will continue to be observed. More cases should be examined in the future with regard to the influence of ring chromosome 8 on mosaic trisomy 8 syndrome.

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