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Trisomy 17 Mosaicism in a Four-year Seven-month-old White Girl: Follow-up Report

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I read with interest the report by Djalali *et al.* (1998) published in this journal.

These authors reported three new cases of true mosaic trisomy 17 diagnosed in amniotic fluid cells. Postnatal chromosome analysis from lymphocytes did not confirm the trisomic cell line. Follow-up studies showed normal psycho-motor development of the children. The longest follow-up was at $4\frac{1}{2}$ years of age in one subject.

I would like to bring attention to our earlier report of a rare case of trisomy 17 (Butler *et al.*, 1996) identified by amniocentesis at 16 weeks of gestation in a fetus from a 23-year-old white mother (gravida III, para I, ab I) with an elevated maternal serum alpha fetoprotein level on two occasions and report follow-up. The amniocentesis results showed a 47,XX,+17 karyotype in 26 cells from 26 colonies from three primary cultures. No fetal anomalies or growth retardation were noted by ultrasonography. A repeat amniocentesis was performed at 19 weeks of gestation and a 46,XX karyotype was seen in six cells from six colonies and a 47,XX,+17 karyotype in 15 cells from 15 colonies. The baby was born by normal vaginal delivery at approximately 40 weeks' gestation weighing 2948 g (20th percentile) and was 50.8 cm long (60th percentile). No minor anomalies were noted and no neonatal problems detected. A cord-blood specimen was collected for chromosome studies and 30 metaphases were analysed and showed a normal 46,XX female chromosome complement. She had a 10-word vocabulary at about one year of age and could stand independently and walk with assistance. She was healthy and on no medication. She had normal growth parameters.

She is now four years seven months of age and is doing well. Her growth parameters have been within the normal range. She required respiratory therapy intermittently between one and two years of age because of asthma which has resolved. Since then she has had no chronic illnesses, allergies or on any medication. She has had no hospitalizations or operations. There have been no seizures or behaviour problems. She began walking at one year of age and has normal co-ordination. She is normally active. She has an unlimited vocabulary and knows her A,B,Cs. She can count, recognize colours and is beginning to print her name. She will start regular kindergarten at the appropriate age. The family history was unremarkable with no changes or additions since the previous report.

Because of the rarity of this cytogenetic condition, we report follow-up on our patient at four years seven months of age and with normal physical and psychomotor development. Our patient with trisomy 17 mosaicism by prenatal diagnosis was reported earlier (Butler *et al.*,

1996) but was not noted by Djalali *et al.* (1998) in their short communication on trisomy 17 mosaicism cases reviewed from the literature.

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