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Antenatal Diagnosis of Partial Trisomy 8 and Partial Monosomy 1: A Case Report

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ABSTRACT A case of prenatally diagnosed partial trisomy 8 and partial monosomy 1 is described. The syndrome is associated with skeletal and cardiac anomalies, as well as hepatic calcification, presented with severe central nervous system malformations. Homogenous and complete trisomy 8 is an extremely rare case. In this case routine karyotyping with (Giemsa-Trypsin) GTG banding for diagnosis was carried out from amniotic fluid at 16 weeks of gestation. Peripheral blood of both parents were karyotyped. Only the father was found to have an abnormal karyotype; reciprocal translocation between chromosome 1p and 8p [46,XY,t(1;8)(p36.1;p21.3)]. The fetal karyotype was, [46,XY,der(1)del(1)(p36.1→pter) dup(8)(p21.3→pter) t(1;8)(p36.1;p21.3)]. This reported case of partial trisomy for this region of chromosome 8 is the first prospectively diagnosed antenatal case in which ultrasound anomalies are noted.