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Phenotypic recognition of maternal (mosaic) and paternal (segmental) isodisomy for chromosome 14 without a Robertsonian translocation. S.P. Yang[1], D.R. Towner[1], M.P. Sherman[1], L.G. Shaffer[2], J.P. Johnson[3]. [1]U.C. Davis Health System, Sacramento, CA, [2]Baylor College of Medicine, Houston, TX, [3]Shodair Children's Hospital, Helena, MT.

We report 2 cases of UPD-14 initially suspected on the basis of clinical rather than cytogenetic clues. The girl with maternal isodisomy presented prenatally with a positive triple marker screen for Down syndrome but a 46,XX karyotype on amniocentesis. Growth retardation, borderline microcephaly, hypotonia and multiple minor anomalies prompted a placental chromosome study that showed non-mosaic trisomy 14. Blood karyotype was normal and UPD-14 was confirmed at 6 weeks. She continues to have hypotonia and delayed motor but normal cognitive skills at 2-1/2 years despite the appearance of streaky hyperpigmentation on all four limbs suggesting low-grade mosaicism for a trisomic cell line.

The boy with paternal isodisomy was found by prenatal ultrasound to have an omphalocele, polyhydramnios and mild skeletal disproportion but chromosomes were 46,XY in amniocytes, lymphocytes and fibroblasts. Major anomalies involving the CNS and cardiovascular system were noted postnatally. The unusual radiographic appearance of his ribs resembled that described in other reports of paternal UPD-14 (Walter, 1996; Cotter,1997). Molecular confirmation was obtained at 7 months but a plugged tracheostomy tube caused severe anoxic brain injury and he remains hospitalized at 10 months. Markers from 14pter through 14q11.2 (D14S740 and D14S1422) are biparental while those from 14q12 (D14S599 and D14S306) through 14qter are uniparental. This implies that paternally imprinted genes on chromosome 14 are located at least 20 cM from the terminal short arm