Sebold C, Roeder E, Zimmerman M, Soileau B, Heard P, Carter E, Schatz M, White WA, Perry B, Reinker K, O'Donnell L, Lancaster J, Li J, Hasi M, Hill A, Pankratz L, Hale DE, Cody JD (2010). Tetrasomy 18p: Report of the molecular and clinical findings of 43 individuals. Am J Med Genet A 152(A): 2164-72.

Based on the reports available in the literature, people with tetrasomy 18p frequently have neonatal feeding problems, growth retardation, microcephaly, strabismus, abnor-malities in muscle tone, scoliosis/kyphosis, and variants on brain MRI. Developmental delays and cognitive impairment are also present. However, this information comes mostly from case reports. The purpose of this paper was to more fully describe the med-ical concerns reported in people with tetrasomy 18p. In addition to completing microar-ray analysis of 43 participants, we completed a thorough review of all the available medi-cal records. We also brought 31 research participants down to San Antonio to complete a series of thorough clinical evaluations. Taken together, the data show us that people with tetrasomy 18p have an increased incidence of neonatal jaundice and respiratory dis-tress; recurrent otitis media; hearing loss; seizures; refractive errors; constipation and gastroesophageal reflux; cryptorchidism; heart defects; and foot anomalies. A minority of participants had hernias, spina bifida, short stature, and failed to respond to growth hormone stimulation testing.