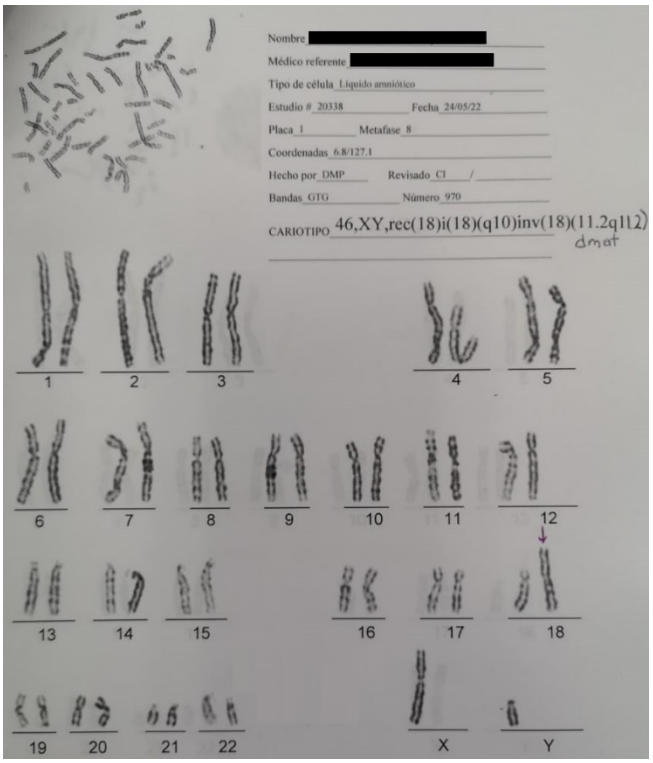
20. **PRENATAL DIAGNOSIS OF A FETUS WITH PARTIAL DUPLICATION AND DELETION OF CHROMOSOME 18 DUE TO MATERNAL PERICENTRIC INVERSION 18.**

Manuel Alejandro Vásquez Salguero[1], Wilmar Saldarriaga Gil[2].
[1] Fourth-year Medical Student. Universidad del Valle, Cali, Colombia.
[2] Head profesor, Universidad del Valle, Cali, Colombia.

https://www.youtube.com/watch?v=0JIMP5FyI7s&t=6549s

INTRODUCTION: The phenotype of structural chromosome 18 mutations is highly heterogenous, clinical manifestations may range from mild to severe, they have been widely studied in the literature, however, there are few cases where two or more mutations are present in the same individual, reports where these alterations are caused by a maternal pericentric inversion and diagnosed prenatally are even rarer. Affected individuals are generally characterized by low birth weight, intellectual disability, heart defects, musculoskeletal abnormalities, craniofacial anomalies, among others. We describe the case of a masculine fetus from a non-consanguineous marriage obtained at week 29 of gestation. 27-year-old mother, G2C1, she started antenatal care at week 11 of pregnancy at the Hospital Universitario del Valle, in Cali, Colombia. Ultrasound at the 11th week of gestation showed a 3.9 mm nuchal translucency (>95p), subsequently at week 18th a new ultrasound found a 6mm nuchal translucency (>95p); suspecting a chromosomal aberration an amniocentesis was carried out at week 18th, a 756-band resolution karyotype reported: chromosome 18 partial trisomy (from 18p.11.2 to the centromere and from 18q11.22 to 18qter), and chromosome 18 partial monosomy (from 18pter to 18p11.2 and from the centromere to 18q11.2). The parents also underwent genetic testing, the karyotype of the mother exhibited a pericentric inversion of the 18 chromosome (46,XX,inv(18)(p11.2q11.2), no abnormalities were found in the father's genetic material. At week 24 of gestation detailed anatomy ultrasound showed left diaphragmatic hernia, multicystic dysplastic kidney, and polyhydramnios. After explaining the diagnosis and providing genetic counseling, the patient requested voluntary interruption of pregnancy as established by the c-355/06 law of the Colombian constitutional court. A 1,180 g fetus was obtained. The structural rearrangement of this case may be explained by an error in maternal meiosis, oogenesis. During meiosis 2 an anomalous disjunction in the chromosome 18 took place, where the two short arms were separated from the long arms, the latter isochromosome was the one that the fetus received, thus, leading to the described mutation. The aim of this case report is to provide to the scientific literature the first case of prenatal diagnosis of a fetus with partial trisomy and partial monosomy of specific bands of the chromosome 1, due to a maternal pericentric inversion, while also highlighting the value of early prenatal diagnosis, in order to make choices regarding voluntary interruption of pregnancy, pregnancy follow up, or planning the delivery method, allowing adequate genetic and reproductive counseling for future pregnancies.

Figure. Karyotype of the Fetus.



Legend: 756-band resolution karyotype depicting a structural mutation in the chromosome 18.

Key words: Chromosome Disorders; Chromosome 18; Trisomy 18q, Partial trisomy, Partial monosomy.