P35 | Cytogenetics & Genomics

Derivative chromosome 7 in a newborn with hypotelorism, cleft palate, agenesis of corpus callosum and semilobar holoprosencephaly

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Cytogenetically visible unbalanced chromosome rearrangements involving the euchromatic regions most often result in severe phenotypic features. Often, it is not possible at microscopic level to distinguish if a chromosomal anomaly involves one or more than one chromosome. In these cases, the parents study is fundamental and is usually the first line of study. We report a female newborn with multiple anomalies. Ultrasonography at 32+6 weeks of gestation revealed moderate ventricular dilatation, microcephaly and intrauterine growth restriction (IUGR). Delivery was at 35 weeks and microcephaly, hypotelorism, complete medium cleft palate with nasal depression, agenesis of the corpus callosum, thalamic fusion and fusion of the lateral ventricles in the frontal region suggestive of semilobar holoprosencephaly (HPE) was observed. Seizures and nistagmus were described since the eighth day. Hypotonia was present. In addition, diabetes insipidus was diagnosed. Sepsis was developed at day 14 followed by death at day 18 in consequence of seizures and respiratory insuficiency. Cytogenetic analysis revealed an abnormal chromosome 7qter as a result of an unbalanced segregation of a maternal reciprocal translocation t(7;19), with breakpoints at 7q36.1 and 19q13.42. The newborn karyotype is 46,XX,der(7)t(7;19)(q36.1;q13.42)mat. The patient presented a partial trisomy of the region 19q13.42→qter and a partial monosomy of the region 7q36.1→7qter. Partial monosomy of chromosome 7qter has been characterized by a wide phenotypic manifestations, but HPE, microcephaly, midface hypoplasia, maxillary anomalies and sacral agenesis are frequently described. However, is not often reported in newborns. Partial trisomy 19q is a rare and severe condition, and has been described associated with low birth weight, growth retardation, microcephaly, seizures, dysmorphic facial features, short neck, clynodactyly, heart malformations, anomalies of the genitor-urinary and gastrointestinal tract. To our knowledge, there is only one previous case of der(7)t(7q;19q)(q36.1;q13.43) described, in a fetus who presented severe sacral agenesis and IUGR. The case herein reported presents some of the most common features of 7q36 partial monosomy and 19q terminal trisomy, although some of them are present in both conditions. The presence of those two imbalances may complicate the final phenotype but the important matter will be the counseling of the couple and to prevent future imbalances in their offspring.