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18p deletion syndrome: foetal phenotype and cytogenetic characterization

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Monosomy for the short arm of chromosome 18 is one of the most common autosomal deletion syndromes, with an estimated incidence of about 1:50000 live-born. The phenotype of this chromosome imbalance is greatly variable, often not evident at birth. In this communication we describe a case prenatally diagnosed of de novo 18p deletion.Amniocentesis was performed at 17 weeks of gestational age on a 39-year-old gravid. The family history was unremarkable and the pregnancy was uncomplicated. At the time of the amniocentesis the amniotic fluid alphafetoprotein level was slightly lower than normal. The pregnancy was terminated at 18 weeks, after the cytogenetic response. At the time the ultrasonographic examination showed reduced biparietal diameter and head circumference, normal limbs, normal thoracic, abdominal and pelvic organs.

The external examination of the aborted foetus showed: weight 380 gr; ipsicephaly and nuchal oedema, no facial dysmorphic signs, normal ears, limbs of normal length, overlapping toes in the right foot. External genitalia were normal. At the autopsic examination the right lung showed four lobes; all the other organs of thorax, abdomen and pelvis were normal.Cytogenetic analysis of cultured amniocytes showed a de novo 18p deletion. FISH studies allowed us to define the foetal karyotype as: 46,XX, del(18)(p10pter).ish del(18)(tel18p-, dim D18Z1).

The phenotypic spectrum of 18p- syndrome may include: round face, ptosis, flat and broad nasal bridge, wide mouth with short upper lip, small mandibles, irregular teeth and dental caries, large, protruding ears, often low and posteriorly rotated, short neck, pectus excavatum, kyphoscoliosis, hands and feet anomalies. The main malformations are holoprosencephaly (HPE), cardiac defects and genital anomalies. Growth retardation, psychomotor delay and mental insufficiency, often mild, are constant symptoms of the syndrome.

The foetal phenotype is inconsistent, but the increased nuchal translucency (INT) seems to be a sonographic sign suggestive of this chromosomal abnormality. This further stress the importance of accurate morphological ultrasonographies, mainly in the second trimester, in order to prevent rare and oligosymptomatic chromosomal syndromes.

Keywords: 18p deletion syndrome