

Case report: A rare mosaicism on chromosome 21

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Abstract/Resumo

Changes in the human karyotype result in alterations with varying degrees in relation to the morphology or intellectuality of the affected individuals. In most cases, genetic alteration can lead to spontaneous abortion. Among the chromosomal anomalies, mosaicism is often found, consisting of more than one type of cell line, derived from a single zygote, that is, mosaicism is a post zygotic phenomenon that can originate germ cells or somatic cells. The purpose of this succinct explanation is to show the cytogenetic diagnosis of the EVS patient (1 year and 9 months old) who presented the following chromosomal mosaicism at 21 chromosomes. The result: 46, XY, del (21) (pter→q11.2::q11.2→qter)[75]/46,XY,+21,der(21;21)(q10;q10)[22]/45,XY,-21[3]. Consequently, proband presents Down's Syndrome, with almond eyes and small ears and low implantation. This patient does not present with cardiopathy or intestinal problems. The karyotype shows that mosaicism is much rarer, since the existence of three different cell lines. 75% proband cells have a partial deletion of the long arm of chromosome 21 in its portion q11.2. A second cell line encompasses 22% of cells analyzed consisting of a Robertsonian translocation derivative between the long arms of chromosomes 21 and the remaining 3% there was a complete deletion of a 21 chromosome yielding a karyotype with 45, XY with only 1 chromosome 21. Thus and result found shows the need for follow-up of the patient to observe their development and changes that may better characterize their clinical conditions.

Support: PROEXT

Keyword/Palavras-chave: Mosaicism; Robertsonian Translocation; Down's syndrome; Complete deletion

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