V Reunião Brasileira de Citogenética e Citogenômica
5th Brazilian Meeting of Cytogenetics and Cytogenomics
30 e 31/Maio & 01 e 02/Junho de 2017

Syndrome del(5)(q14~23),der(10)(p12p15)?add(10)(q26): Case report and literature review

Conceição, E.V.[1]; Minamihara, A.S.[2]; Barroso, M.E.L.[3]; Paiva, W.J.M.[4]

Abstract/Resumo

Structural chromosomal mutations are changes that do not modify the number of chromosomes in the cell, but they determine the appearance of abnormal chromosomes. The aim of this study is to report the clinical and cytogenetic history of a patient carrier of a structural mutation with karyotype 46,XY,del(5)(q14~23),der(10)(p12p15)?add(10)(q26) attended by the Genetic Counseling Service of the State University of Londrina. The patient (ABT) is male, had 1 year and 9 months on the day of the examination, normally born, full term (38 weeks) with Psychomotor Development Retardation (PDR) and without other complications. At birth, ABT's mother was 25 years old and his father was 31 years old. The x-ray, urine analysis, pre- and postnatal ultrasonography, parasitological, ophthalmologic, gastrointestinal and neurological exams do not reveal changes. At the time of cytogenetic examination, the patient underwent surgery for the treatment of esophagogastric reflux and inguinal hernia. Currently, ABT follows up with Pediatrician, Social Worker, Physiotherapist, Speech Therapist, Neuropediatrician, Nutritionist and Ophthalmologist. These findings, particularly PDR, were consistent with the rare syndrome of the 10q + chromosome, since only ten cases of trisomy for the distal segment of the major arm of chromosome 10 (10q +) were described. Excluding the PDR, other characteristics were not found in the trial. Structural chromosomal syndromes represent an opportunity to understand clinical phenotypes and become important in gene mapping, especially when associated with molecular genetic techniques, the next step in diagnostic genetic research. In this way, the patient follows the treatment of the symptomatology.

Keyword/Palavras-chave: Structural chromosomal mutations; Psychomotor development retardation; Syndrome of the 10q + chromosome

1 Medical Student at UEL, Londrina-PR, *emersoncalixto83@gmail.com*
2 Student of Biological Sciences at UEL, Londrina-PR
3 Laboratory Technique at UEL, Londrina-PR
4 Professor at UEL, Londrina-PR