332.00

DE NOVO TRISOMY -8 MOSAICISM-A FIVE YEARS FOLLOW UP

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The trisomy 8 mosaicism is extremely variable in its phenotypic and cytogenetic expression.
The clinical features include mental retardation, dysmorphy of the face, skeletal anomalies (particulary vertebral), congenital hearth defect and malformationes of the kidney. The patients are usualy not recognizible at the birth with mayor anomalies, but with dysmorphy of the face and deep longitudinal palmar and plantar furrows. Mental retardation, infections, immunodeficiency and malignancies are described.
Here we present a boy with trisomy 8 mosaicsm. We are discussing about correlation between clinical and laboratory findings. His karyotype at the begining was: 47,XY,+8 (62%)/46,XY (38%) and mosaicism for trisomy 8 was detected in all tissue examined in the same proportions. After five years we found decreasing of the trisomic cells in the proportins of 50% in peripheral blood lymphocytes. This indicate the mitotic instability of aneuploid cells during growth