



EUROPAEDIATRICS•2003

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This CD contains the abstracts of the Second European Congress in Paediatrics "Europaedi Prague, Czech Republic, October 19-22, 2003.

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DE NOVO TRISOMY -8 MOSAICISM-A FIVE YEARS FOLLOW UP**V. Culic¹, B. Lozic¹, S. Culic², B. Resic³, D. Primorac⁴, R. Lasan⁵, R. Martinic⁶***¹Paediatrics Medical Genetics ²Paediatrics Hematology ³Paediatrics Developmental Neurology**⁴Laboratory for Molecular Genetics ⁵Laboratory for Molecular Cytogenetics**⁶Laboratory for Tissue Typing, Clinical Hospital, Split, Croatia**vida.culic@st.hinet.hr*

The trisomy 8 mosaicism is extremely variable in its phenotypic and cytogenetic expression. The clinical features include mental retardation, dysmorphism of the face, skeletal anomalies (particularly vertebral), congenital heart defect and malformations of the kidney. The patients are usually not recognizable at the birth with major anomalies, but with dysmorphism 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 mosaicism. We are discussing about correlation between clinical and laboratory findings. His karyotype at the beginning 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 proportions of 50% in peripheral blood lymphocytes. This indicates the mitotic instability of aneuploid cells during growth.