

A Down-szindróma genetikai természetete

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Abstract: Down syndrome is a result of trisomy of chromosome 21, but about 4-5% of cases are caused by a translocation. The most common translocation is of chromosome 21 to chromosome 14 (or less commonly to chromosome 13,21,22 or another chromosome). Approximately 1-2% of patients have mosaicism with two cell populations, one normal and one trisomic for chromosome 21. Patients with Down syndrome are mentally retarded and have highly characteristic appearance.

Keywords: trisomy, translocation, mosaicism, nondisjunction, advanced maternal and paternal age.