

Down syndrome (DS): Studies focusing on risk factors other than advanced maternal age

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It is well known that the risk of aneuploid conception increases with advancing maternal age. However, the majority of individuals with trisomy 21 have been conceived by young couples. This presentation is a synopsis of the author's published and most recent studies on age-independent factors favoring to chromosome 21 nondisjunction. Our data give evidence for: low coital rates in young parents of DS offspring; an implication of maternal hormonal imbalance and hypo function; an effect of habitation at poor living conditions complicated by unfavorable neighborhood (communal flats and hostels); an effect of some geographically fixed exposures other than radon; the majority of the mosaic parents were conceived as trisomics; male excess in non mosaic DS individuals as the result of intrauterine selection against females; female predominance among both mosaic DS individuals and mosaic parents due to sex-specific instability of chromosome pericentromeric regions. Our data failed to support: advanced grandmaternal age as a risk factor for young parents; an effect of economic crisis due to social transition, accompanied by about 3-fold reduction in birth rate, on both incidence and infant mortality of DS individuals; genetic predisposition to occurrence of double aneuploidies involving chromosome 21 and sex chromosomes; interchromosomal effect in parent carriers of balanced non-contributing chromosome rearrangement, instead, one may suggest a post fertilization effect of paternal rearrangement on segregation of maternal chromosomes. Above all, though irrelevant to chromosome 21 nondisjunction etiology, there is an intriguing finding of strong female excess among false-positive diagnosis of DS, particularly in newborns and infants.

Biography

Natalia V. Kovaleva received her PhD in Genetics from Leningrad Research Institute of Animal Breeding and Genetics in 1984. She is a Senior Researcher at Scientific Research Centre, St. Petersburg State Pediatric Medical Academy, chairperson of the Board of St. Petersburg Society of Medical Genetics (since 2004) and member of the Board of All-Russian Society of Medical Genetics (since 2010). She has published more than 50 papers in reputed journals. She had founded and has been running St. Petersburg Down Syndrome Register containing data on about 3,000 patients/fetuses with trisomy 21 and their parents since 1970.

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