Cytogenetic analysis of 1372 intellectually disabled children

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Objective
To analyse the relationship between mental retardation and chromosomal abnormalities in children, and evaluate the significance of prenatal screening and diagnosis in preventing the birth of intellectually disabled children.

Methods
In 1372 children under 14 years old the peripheral blood was cultured through routine method and chromosome G band.

Results
In 1018 (74.20%) cases out of 1372 intellectually disabled children, the karyotype was abnormal. Chromosome aneuploidy, mainly trisomy 21 (Down syndrome) was found in 953 cases, representing 93.61%. 42 cases (44.13%) of unbalanced abnormal chromosome structure were detected. There were 6 cases of balanced chromosomal structural abnormalities (0.59%), as well as 1 case of extra sex chromosome (47, XXY) and 1 case of abnormal sex chromosome structure (46, X, der(Y)). 10 cases of marker chromosome karyotype, representing 0.98%, were also detected.

Conclusion
Intellectual disability in children is closely related to abnormal karyotypes. The prenatal diagnosis helps identifying these cases and is an efficient method for preventing the birth of intellectually disabled children.