Abstract

Microdeletion syndromes are complex diseases caused by loss of genetic information resulting from cryptic deletions which are smaller than 5 Mb. They are cause a large number of phenotypic features. Most common are developmental and mental retardations, various physical defects and abnormalities or behavior problems. It has been shown, that in some cases plays a role parental origin of affected chromosome in microdeletion syndrome. In Angelman, Prader-Willi and Beckwith-Wiedemann syndromes is unequal disability of chromosomes caused by genomic imprinting. The reasons for dominance disability of one parental chromosome in Cri du chat syndrome, monosomy 1p36 and Phelan-McDermid syndrome are different and the effect of genomic imprinting has not been confirmed.

Key words: microdeletion, microdeletion syndromes, methylation, genomic imprinting