CLINICAL AND CYTOGENETIC POLYMORPHISM IN KLINEFELTER SYNDROME

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Introduction. Klinefelter's syndrome, the most common chromosomal abnormality with an incidence of 1 : 500–700 newborns with a male phenotype, is characterized by the additional presence of one (rarely several) X chromosome in boys.

The aim. Is to study the peculiarities of the clinical and cytogenetic polymorphism of Klinefelter Syndrome in different periods of ontogenetic development for an early diagnosis of children.

Material and methods. The study was performed on 73 children of pediatric age, selected during medical genetic counseling in the Center for Reproductive Health and Medical Genetics, having the following phenotypic selection criteria: developmental anomalies of the external genitalia — peno-scrotal hypospadias, micropenis, small testes, cryptorchidism, cranio-facial dysmorphism, waist high and disproportionate, hypogonadism, gynecomastia, mental retardation, psychosocial problems.

Results. Klinefelter's syndrome was confirmed cytogenetically in 32 patients. The most common cytogenetic variant diagnosed was homogeneous free trisomy 47,XXY (28 cases — 87.5%), followed by mosaic form (47,XXY/46,XY: 2 cases — 6.2%), polysomy X (variant 48,XXYY: 1 case — 3.1% and pentasomy — 49,XXXXY: 1 case — 3.1%). Most patients with variant 47,XXY classic and mosaic form showed mild to moderate mental retardation, language disorders with cognitive-verbal retardation, slow motor development, coordination disorders, immature behavior. In variants 48,XXYY and 49,XXXXY, moderate to severe mental retardation, severe cognitive-verbal retardation, severe behavioral problems. Most patients with Klinefelter Syndrome had been diagnosed in puberty (22 cases — 68.7%), 6 patients (18.7%) were diagnosed prepubertal, and only 4 patients (12.5%) were diagnosed during early childhood.

Conclusions: Early recognition of Klinefelter's syndrome, with cytogenetic and phenotypic heterogeneity, allows the initiation of the correct treatment, the prevention of complications and the minimization of the negative psycho-social impact. The cytogenetic variant of Klinefelter syndrome correlates with the severity of the clinical picture, being directly proportional to the number of supernumerary X chromosomes.