Neurogenetic aspects in men with Klinefelter's syndrome

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Abstract

Background: Klinefelter's syndrome (KS) is the presence of one or more supernumerary X chromosomes. The aim was the investigation of the cytogenetic variant in men with KS, for the assessment of neurological phenotypes.
Material and methods: Were investigated 98 men with infertility, having as selection criteria, lack of sperm in the ejaculate, elevated values of Follicle-stimulating hormone (FSH), Luteinizing Hormone (LH), and the following phenotypic aspects: small testes, hypogonadism, cryptorchidism, waist high and disproportionate, gynecomastia, mental retardation, psychosocial problems. Karyotyping was performed according to standard methods G-banding.
Results: The most common cytogenetic variant diagnosed in 25 (25.5%; [95 CI 21.1 – 29.9], p = 0.05) patients with SK was homogeneous free trisomy 47, XXY (22 cases – 88%), followed by: mosaic form (47, XXY / 46, XY: 1 case), polysomies X – Y variants (48, XXYY and 49, XXXXY: 2 cases). In the patients with variant 47, XXY the classical and mosaic forms showed a mild to moderate mental retardation (36.0%; [95 CI 26.4 – 45.6], p = 0.05), language disorders with cognitive-verbal retardation (48.0%; [95 CI 38.01 – 57.99], p = 0.05), slow motor development (20.0%; [95 CI 12.0 – 28.0], p = 0.05), coordination disorders (8.0%; [95 CI 2.57 – 13.43], p = 0.05), immature behavior (60.0%; [95 CI 50.2 – 69.8], p = 0.05). In patients with variants 48, XXYY and 49, XXXXY, moderate to severe mental retardation (50.0%; [95 CI 14.64 – 85.36], p = 0.05), severe cognitive-verbal retardation (50.0%; [95 CI 14.64 – 85.36], p = 0.05), behavioral problems and life-threatening problems were found in 100%.
Conclusions: The cytogenetic variant of KS depends on the number of supernumerary X chromosomes, being determinant in the characteristic of neurological phenotypes.
Key words: Klinefelter's syndrome, cytogenetic, neurologic, phenotype.

Correlation between neurological impairment and liver status in Wilson's disease

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Abstract

Background: The most widely recognized aspect of the neuro-hepatic relation is hepatic encephalopathy, in which neurotransmission in the brain is altered. Of course, there are many conditions that affect both the liver and the nervous system, Wilson's disease being one of the best known. The aim was to characterize the neurological manifestations of Wilson's diseases in terms of symptom type and degree of neurological impairment and correlate these features with degree of abnormalities in copper metabolism, and hepatic status.
Material and methods: 15 patients diagnosed with Wilson's disease were characterized by examination in terms of symptoms including consciousness, activities of daily living as reported by the patient. The neurological manifestations were analyzed in relation to copper abnormalities and liver status.
Results: Most patients (52.9%) exhibited tremor and ataxia, whereas 9.3% were dystonic, and 7.3% had Parkinsonism. Discrete signs were observed in 19.6% of patients. A positive correlation between neurological impairment and higher level of free cooper was observed (Pearson $r = 0.71$). Poor correlation was identified between neurological impairment and hepatic disturbances.
Conclusions: The neurological manifestations of Wilson's disease did not appear to be correlated with hepatic status. These results draw our attention to the symptomatic variability of Wilson's disease, and an individualized approach to each patient is essential.
Key words: Wilson's disease, hepatic copper, neurological status.