

Introduction: Turner syndrome (TS), in which there is loss of all or part of one sex chromosome, occurs in one in 2500 live-born females and is associated with characteristic clinical and physiologic abnormalities -short stature and gonadal failure.. Gonadal function is also clinically important, ranging from the onset of spontaneous puberty and the potential for fertility to complete gonadal failure. Klinefelter syndrome (KS) affects males carrying an additional X chromosome, which results in male hypogonadism, androgen deficiency, and impaired spermatogenesis. Some patients may exhibit all of the classic signs of this disorder, including gynecomastia, small testes, sparse body hair, tallness and infertility.

Purpose and objectives: To evidentiates the growth hormone treatment effect in increasing height in TS and the testosterone replacement therapy effect in virilisation in KS.

Materials and methods: Our study includes 5 cases diagnosed with Turner syndrome and 3 cases with Klinefelter syndrome between 2005-2011 in Iasi Medical Genetics Center in order to illustrate some variants and to show the cytogenetic complexity of these syndromes which reflects in the clinically diverse presentations. Cytogenetic diagnosis was performed using peripheral lymphocytes with G banding and Fish analysis.

Results and discussion: Of the 5 cases that had been diagnosed with Turner Syndrome, all of them had various skeletal malformations ranging from the shortening of the fourth metacarpal to dental abnormalities, face malformations and hypostature while the karyotypes were slightly different between the 5 cases analyzed. Furthermore, one case (karyotype 45,X/46,X,r(X)(p22.1q24)[24]/[23]) had a congenital unilateral ovary which added to the complexity of the clinical approach. We also found one case (karyotype 46, XX/45,X[41][7]) that had the characteristic sausage-like appearance of the toes.

Of the 3 cases diagnosed with Klinefelter syndrome, 2 cases had distinctive intellectual and learning disabilities (karyotypes 48,XXY[96]/[3] and 48,XXYY/47,XYY), while in the other case (karyotype-47,XXY(38)) hypogonadism, which led to gynecomastia and late puberty, which in turn stood as the basis for psychosocial problems, represented the main features.

Conclusion: Our study shows the cytogenetic complexity of Turner and Klinefelter syndromes which is reflected on the clinical features of the patients outlining the importance of karyotyping in these plurimorphative syndromes.

Keywords: genetic syndrome, karyotype, hypogonadism

DIFFERENCES IN FROG SKELETAL AND CARDIAC MUSCLE EXCITATION

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Introduction: The problem of muscle excitation is an important component of research in experimental physiology as well as in clinical sciences such as cardiology and sports medicine. When trying to determine intercellular interactions of the excitation transmission, there were restrictions of methodical nature. We propose a new methodological approach to assess the distribution of action potentials in skeletal and cardiac muscles of frogs.

Methods and Results: For action potentials recording we used polytrode (multichannel conical microelectrode), sharpened as a pencil. With this treatment the contact areas are located in one zone close to each other at a fixed distance.

The polytrode was placed into the intercellular space of gastrocnemius muscle of a frog and potentials of several adjacent muscle fibers were recorded.

We recorded spreading of excitation in the gastrocnemius muscles during tetanic contraction and during the rest period. It was found that tetanus is accompanied by the rhythmic action potentials that were recorded by all channels of the polytrode. During the rest time a tonic contraction takes place, when single action potentials are observed, enveloping only individual muscle fibers without being distributed to the neighboring fibers. Such contractions were recorded as a series of potentials at individual contact sites of the polytrode.

After recording the action potentials of skeletal muscle we have placed the polytrode into the cardiac muscle tissue. The peculiarity of the heart is a solitary contraction when short potential covers sequentially all fibers and it was recorded on the all the channels simultaneously as one solid “wave”.

Conclusion: Our research shows that the excitation of skeletal muscle demonstrated individual potentials from individual muscle fibers. It looks very similar to a myogram of skeletal muscle during the titanic contraction. In the cardiac muscle due to the peculiarities of its structure and ability to transfer the action potential from one cardiomyocyte to another the oscillogram looks like summation of excitations of individual fibers with a very small interval.

Key words: muscle excitation, tetanic contraction, skeletal muscle, cardiac muscle, frog.

COUPLED SPIKE ACTIVITY IN MICROPOPULATIONS OF THE CORTEX NEURONS

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Introduction: As it is known, in many CNS structures, neurons, which are spatially close to each other, form micropopulations. These neurons are characterized not only by neighboring spatial localization but also by the existence of close functional synaptic connections between members of such population. The phenomenon of association of cortical neurons in the so-called columns or rather similar groupings (barrels, etc.) is widely known. But the functional relations between members of such micro populations remain little studied. In our research we recorded rigid coupling of the impulse activity generated by two spatially close cortical neurons that were observed in many cases.

Methods and Results: Using eight-channel metal microelectrodes (diameter of a separate channel 12 μm), we extracellularly recorded the impulse activity of 186 single neurons or their small groups (usually, pairs) localized in the motor cortex of rats anesthetized with ketamine. In 60 cases (32.3%), APs (action potentials) of two single neurons were generated in a parallel manner and demonstrated fixed time relations between each other. This is interpreted as being a result of excitation of two neighboring functionally connected (coupled) cells. These AP pairs could be recorded via one and the same or two neighboring microelectrode channels. Second AP in the pair was elicited exclusively in the case where an AP was preliminarily generated by another neuron, while APs of the latter in some cases could arrive independently. Therefore, “leading” and “accompanying” cells could be identified in such neuronal pairs. The coupling coefficient in the generation of APs by an accompanying unit with respect to APs generated by a leading cell was close to 100%, without dependence on the discharge frequency in the latter. Intervals between APs of two neurons in different coupled pairs varied from about 1.0 to 22-23 msec.