

## Uncommon pain syndrome in a child – case report.

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We present a case of a 12 years old boy suffering from pain concerning his right lower extremity in whose pathogenesis a significant role is played by some congenital sensory disorders. It is a patient with a negative history towards genetically determined diseases, born at term with congenital contracture of the right hip joint and the right knee joint. Besides these contractures, physical examination shows a cutaneous lesions – the whole right limb is covered with a light brown patch with numerous small dark brown spots. Since the earliest months of the boy's life a different sensitivity to touch in the right leg was observed. The patient himself says that as long as he can remember he has had suffering from pain in the right leg and therefore he has been trying to protect it from any injury or even touch.

Because of the congenital contracture of the right hip the boy was operated on three times, respectively at the ages of 2, 6 and 9. The third operation was complicated by a total cutting of the femoral artery. Since that time the pain has become more intensive and, at present, it refers mainly to the right thigh. So far the child has gone through detailed radiological diagnostics (X-ray of hip joints, CT and MRI of the head and the spinal cord), electrophysiological examination (evaluation of the speed of conduction in motor and sensory fibres), histological examination of the skin and muscle segment of the right thigh, thermovisual examination as well as qualitative assessment of sensation. Some anomalies were observed in the last mentioned examination: thermal allodynia for the temperature of 40°C and mechanical static allodynia referring mainly to the right thigh.

Pharmacological treatment applied: oxcarbamazepine – no effects, gabapentine – about 20% reduction of pain. The boy's parents refuse their consent to any invasive methods of treatment. The boy undergoes rehabilitation all the time and is under constant psychological care.

Although the patient has been consulted by several experienced neurologists and pain treatment specialists, the hereditary disease responsible for his pain has not been identified. In differential diagnosis we took under consideration some congenital syndromes that may be accompanied by pain, e.g. a large group of hereditary neuropathies and facomatoses. We have searched the worldwide databases, but we have failed to find a similar syndrome. Since the symptoms refer to the nervous system and the skin, we suppose that our patient suffers from a form of facomatosis, not to be found in the literature, and the pain is the result of overlapping of some congenital sensory disorders and iatrogenic lesion of the right femoral nerve.