

Uncommon pain syndrome in a child – a case report

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Case report - anamnesis

A twelve-year-old boy with a negative history of screening for genetic diseases, born at term with a congenital contracture of the right hip joint and the right knee joint.

Besides these contractures, physical examination reveals the presence of cutaneous lesions – the entire right lower extremity together with the buttock is covered with a light brown pigmentary spot with numerous, small dark brown spots of the same nature.

The cutaneous lesions have been present there since birth and in the earliest years of the boy's life their number increased.







Case report - anamnesis

According to the boy's parents, from the very beginning they observed abnormal sensitivity of the right lower extremity to touch stimuli – during nursing activities the child was restless when the activities involved the right lower extremity.

When the boy grew up and started walking by himself, it became clear that he tried to protect his right leg against touch or injury.

According to the boy himself, as long as he can remember, his right leg has been causing trouble, touching it has been painful and because of this, he has been very careful while dressing and washing and has always been trying to protect his right leg against any possible injury.

Case report – anamnesis

Because of the congenital contracture of the right hip joint, the boy was operated on three times – at the age of 2, 6 and 9 respectively. The third operation was complicated by a total dissection of the right femoral artery, that was reunited by means of a 4 cm insert.

Since that time, in the boy's opinion, his pain has intensified and refers mainly to the right thigh.

Case report – diagnostics

- X-ray of the hip joints and pelvis – normal bone structures
- CT and MRI of the brain – regular picture
- MRI of the backbone structures and the spinal cord in the Th7-S4 section – also regular picture
- In electrophysiological examination the values of conduction in motor and sensory fibres of peripheral nerves were normal
- No pathological picture typical of hereditary neuropathies in the histopathological examination of the muscle segment from the region of the right thigh
- No changes typical of facomatoses in ophthalmological examination
- No indications for surgical treatment – the patient was consulted by a vascular surgeon and by an orthopedic surgeon.

Case report – diagnostics

- No differences between both lower extremities in thermovisual examination – it made possible to rule out ischemia as a factor responsible for pain in the right leg and the participation of the sympathetic system in the pathogenesis of the boy's pain
- Qualitative sensory testing performed by means of von Frey's filaments and rolls for testing cold and hot allodynia showed thermal allodynia for the temperature of 40°C as well as mechanical static allodynia.

Case report - treatment

1. Pharmacotherapy:

- Oxcarbamazepine – with no effects
- Gabapentine – about 20% pain relief

2. Rehabilitation

3. Psychotherapy

- ## 4. The boy's parents refuse to consent to any invasive methods of treatment.

Case report - discussion

- In our opinion, the hyperalgesia and allodynia observed in our patient result from an overlapping of some congenital sensory disorders and the iatrogenic lesion of the right femoral nerve
- But one question remains open: what underlying disease causes the congenital sensory disorders?

Discussion – differential diagnosis

One of a group of hereditary motor and sensory neuropathies (HMSN) – doubtful, because characteristic for HMSN is:

- symmetrical paresis and muscular atrophy most often referring, at least in the first phase of the disease, to the lower extremities
- sensory disorders also usually have a symmetrical distribution
- retardation of nerve fibre conductance in electrophysiological examination
- in biopsy some demyelination changes as well as nerve fibres loss on a different scale
- often defects of the osteoarticular system, usually scoliosis or hollow feet

Discussion – differential diagnosis

Facomatoses – a large (more than 50 entities) group of genetic diseases referring to the skin and nervous system. The most frequent of them is neurofibromatosis (NF) type 1:

- cafe-au-lait spot is a characteristic change present since birth in nearly all patients with NF type 1; in children may be the only symptom of NF type 1
- another very characteristic feature for NF type 1 is the presence of neurofibromas; if they appear within the nerve roots, peripheral or autonomic nerves, they may elicit pain by applying pressure to the nerve structures – we have not found any neurofibromas in our patient
- according to the WHO criteria from 1997 to diagnose NF type 1 it is necessary to meet at least 2 out of 7 strictly determined criteria
 - so we cannot diagnose this type of facomatosis in our patient
- clinical symptoms found in our patient do not correspond to the picture of the other types of facomatoses either

Discussion

Maybe another, not described so far
type of facomatosis ??????



Thank you for your attention.