Neuropathic arthropathy of the knee associated with an intra-articular neurofibroma in a child

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We describe a five-year-old child with neurofibromatosis type I who developed a Charcot knee. Infiltration of the joint by tissue associated with the disease caused damage to the proprioceptive mechanism and resulted in severe joint instability, accelerated destruction and development of neuropathic arthropathy.

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Neurofibromatosis is a genetically transmitted disorder characterised by abnormalities of the skin, nervous tissue and bone. These may include café-au-lait spots, neurofibromas, Lisch nodules, optic gliomas, osseous lesions, macrocephaly, short stature and mental retardation. Pseudarthrosis of the tibia and the ulna, dystrophic kyphoscoliosis, spinal manifestations, protrusio acetabuli, dislocation of the hip, hypertrophy of a limb, atlantoaxial dislocation and tendon ruptures may also occur.1-16

Neuropathic arthropathy with progressive destruction of weight-bearing joints was first described by Charcot in 1868.17 Many causes have been suggested including tabes dorsalis, injury of the spinal cord and peripheral nerves, syringomyelia, diabetes mellitus, congenital insensitivity to pain, amyloidosis, leprosy, myelomeningocele and nutritional deficiency.

Charcot joints are rare in children but may be found in association with neurological disorders such as congenital insensitivity to pain, peripheral nerve injuries, diabetic neuropathy and chronic diseases of the spinal cord which lead to sensory disturbances of the limb.17-25

We describe a child who had neurofibromatosis complicated by the development of a Charcot knee due to an intra-articular neurofibroma.

Case report

A 5-year-old girl was seen because she had a limp and painless swelling of the left knee. Her mother and three of her four siblings had neurofibromatosis type I. The child had had several transient episodes of swelling of the knee during the past year accompanied by limping, which had all settled.

Clinical examination showed multiple café-au-lait spots and a mild lumbar scoliosis. The left leg was 2.5 cm shorter than the right. The left knee showed a large effusion, but the range of movement was normal, although with severe anteroposterior instability. There was no tenderness and all laboratory blood tests were normal. Plain radiographs showed severe degenerative changes with marked irregularity of the joint surfaces (Fig. 1).

Tc bone scanning showed a diffuse increase in uptake in the left knee. Electrophysiological studies of the legs were normal. MRI showed infiltration of the knee with abnormal tissue extending posteriorly into the popliteal fossa and invading the anterior cruciate ligament (ACL), the posterior cruciate ligament and the lateral meniscus (Fig. 2).

At arthroscopy, fibrofatty tissue was seen in the intercondylar notch and the ACL could not be identified. The articular surfaces of both tibia and femur showed severe degeneration and the lateral meniscus was detached anteriorly. This was reattached with sutures and an excisional biopsy was performed. Histological examination of this tissue showed evidence of neurofibromatosis with involvement of synovial tissue (Fig. 3). The immunohistochemical stain for S-100 protein was positive in these cells (Fig. 4).

The knee remained unstable and a knee brace was applied.

Discussion

Neurofibromatosis rarely affects a joint and infiltration of the knee with pathological tissue has not previously been reported. In this case the disease had affected the ACL, the
nerve fibres of the synovial tissue and the longitudinal growth of both tibia and femur causing discrepancy in length and severe instability of the joint. Detachment of the lateral meniscus occurred, and there were early osteoarthritic changes due to complete absence of a sense of pain in the knee.

Infiltration of a joint and its structures by tissue associated with neurofibromatosis has been described in association with dislocation of the hip in children.\textsuperscript{8,14-16} This was accompanied by bony changes including narrowing of the neck of the femur, acetabular dysplasia, erosion of the acetabulum and deformities of the pubis and ischium, all caused by the invasion of neurofibromatosis tissue. McCann et al\textsuperscript{18} have described an adult male with neuropathic arthropathy of the knee due to involvement of a spinal nerve by neurofibromatosis leading to an insensitive knee.

In our patient there was a slow, progressive development of neuropathic arthropathy caused by destruction of the neural proprioceptive defence mechanisms in the knee.

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References


