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Case Report Open Access

Pacydermoperiostosis Associated With Pigmented Villonodular Synovitis

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Background

Pachydermoperiostosis is a rare genetic disorder with autosomal dominant or autosomal recessive transmission and variable expression known also as primary hypertrophic osteoarthropathy and Touraine-Solente-Gole syndrome [1-3]. The proposed mechanism of the disease is the increase of seric levels of Prostaglandin E2 (PGE2) produced by inactivating mutations of Hydroxy-Prostaglandin Dehydrogenase (HGPD) enzyme (responsible for PGE2 degradation) or of SLCO2A1 (a PGE2 transporter) [1,2,4]. Increased seric levels of PGE2 are inducing various metabolic modifications of skin, bones, joints and blood vessels. Overproduction of Vascular Endothelial Growth Factor (VEGF) and / or Platelet -Derived Growth Factor (PDGF) induced by the increased levels of PGE2 could play also a significant pathogenic role [1,4]. Alcohol could also contribute to the apparition of the disease [1,4]. The clinical manifestations are pachyderma thickened facial skin with facial coarsening, acropachia - digital clubbing of hands and feet, periostosis, excessive sweating, arthritis, symmetrical enlargement of forearms and legs [1-5]. The male to female ratio is 7: 1 and the disease is usually begins in childhood or adolescence and progresses slowly for 5 - 20 years before stabilizing [1]. The clinical presentation could be complete with skin and musculoskeletal manifestations or incomplete. The laboratory tests are usually normal except for the modifications induced by concomitant diseases. Radiographic examination shows sub periosteal bone formation, cortical thickening, enlargement of phalanges, metacarpal and sometimes of the ulna and radius [1,4].

The diagnostic criteria for pachydermoperiostosis are:

Major criteria - pachyderma, periostosis and digital clubbing

Minor criteria - hyperhydrosis, arthralgia, arthritis, symmetrical (columnar) enlargement of forearms and legs, edema [1,6]. The differential diagnosis should include acromegaly (increased IGF and GH), pachydermoperiostosis secondary to malignanacies, secondary hypertrophic osteoarthropathy (development in adulthood, associated with severe chronic pulmonary diseases, pulmonary malignancies, cyanotic heart diseases) and inflammatory rheumatic diseases [1-5]. The treatment consists usually of long term use of non-steroidal anti-inflammatory drugs, joint fluid aspiration, intraarticular glucocorticoids. There are some reports of use of Colchicine, Sulfasalazine, Methotrexate, and TNF alpha blockers. Intravenous pamidronate or zoledronate could improve musculoskeletal symptoms. Surgical interventions could be indicated for skin problems and also for joint involvement [1-5]. Refractory synovitis is rarely reported in pachydermoperiostosis [7]. Pigmented Villonodular Synovitis (PVNS) is a rare, usually benign, villous or nodular synovial proliferation of joints, tendons and bursae [8-11]. The pigmentation is generated by the accumulation of hemosiderin in the synovial proliferation [10,11].

According to World Health Organization, PVNS is considered a tenosynovial giant- cell tumor and is classified as diffuse type giant cell tumor, with destructive potential and involving a joint and localized giant - cell tumor, involving tendons, bursae or joints [9-11]. The pigmented villonodular tenosynovitis (giant - cell tenosynovitis) is the most frequent manifestation of the disease [9-11]. The knee is the most frequent affected joint, followed by the hip, and then some others joints as elbow, shoulder and ankle [8-11]. The etiology is uncertain, but mutations in the Colony - Stimulating Factor 1 (CSF-1) gene with overexpression of CSF-1 have been described in some cases of PVNS [9]. Joint pain, chronic swelling and limitation of the joint mobility are the most common clinical manifestations and because of non-specificity of signs and symptoms there is usually a delay in diagnosis [8-11]. The laboratory tests are normal. The radiographic examination is normal in the early phases but later it shows joint swelling, bone erosions and subcondral cysts [8-11]. Computed Tomography (CT) shows synovial thickening and bone erosions. The Magnetic Resonance Imaging (MRI) is particularly helpful and very specific. MRI reveals synovial thickening with low to intermediate signal on T1 - weighted images and low signal in T2 - weighted images with enlargement of low signal intensity areas in the gradient echo images ("blooming effect") very specific for the presence of hemosiderin in the synovial tissue [9-11]. The most effective treatment is synovectomy [8-11]. Radiation therapy, external or intra articular, is also used for PVNS [8-11].

Case Report

We present a case of a 45 years old male, admitted in an orthopedic department for pain and swelling of the right knee. The patient had a synovectomy of the left knee 15 years ago for chronic resistant synovitis. In the last 3 years the patient had repeated joint fluid aspiration of the right knee. At the clinical examination the patient had typical modifications of pachydermoperiostosis with coarsened facial features and hand and feet digital clubbing (Figures 1 and 2). The right knee had important swelling with very limited flexion (30 degrees) and normal extension (Figures 3 and 4). The X-ray examination revealed periosteal bone production of the femur, tibia and patella, patellar bone resorption and osteoarthritic modifications of the knee. The MRI examination showed a hypertrophic synovial tissue with villous and nodular protrusions and a large amount of fluid (Figures 5-7).

The orthopedic evaluation suggested an open synovectomy of the right knee. During surgical intervention, an important synovial hypertrophy involving the whole synovium was found with brown discoloration suggestive for hemosiderin deposits, with bone invasion and dystrophic modifications of the right quadriceps muscle and tendon. After surgery the patient had important improvement of pain and mobility of the knee (improved flexion from 30 degrees to 100 degrees). The pathologic examination of the removed synovial tissue (Figure 8) showed pigmented villonodular synovitis. The patient had a favorable evolution and after 1 year he has mild pain and swelling of

the right knee with normal extension and improved flexion (around 120 degrees).

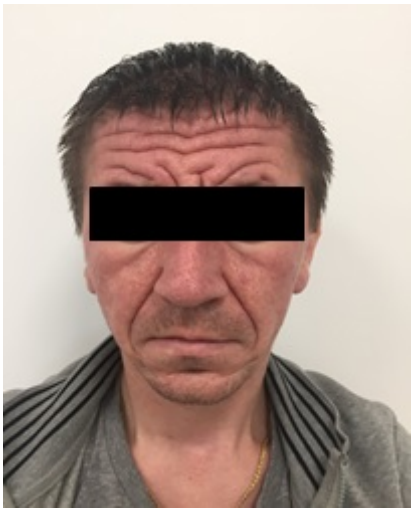

Figure 1: Facial aspect of patient.

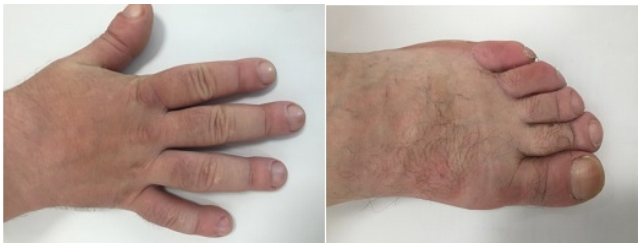

Figure 2: Aspect of hands and feet of the patient.

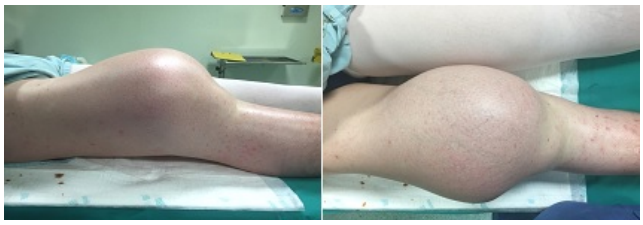

Figure 3: Swelling in right knee with limited flexion and normal extension.

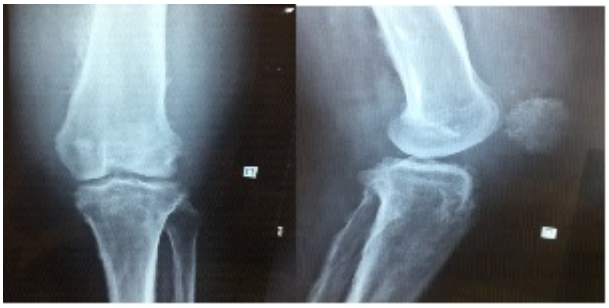

Figure 4: X-ray image of Swelling in right knee with limited flexion and normal extension.

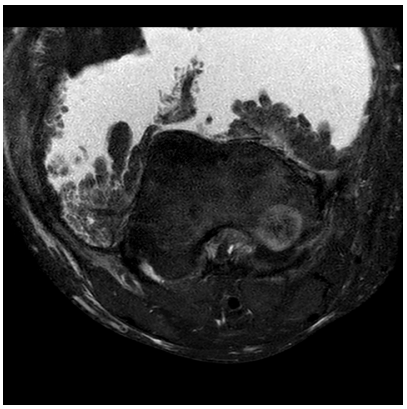

Figure 5: PDW SPAIR axial view showing synovial hypertrophy with villous and nodular protrusions and a large amount of fluid.

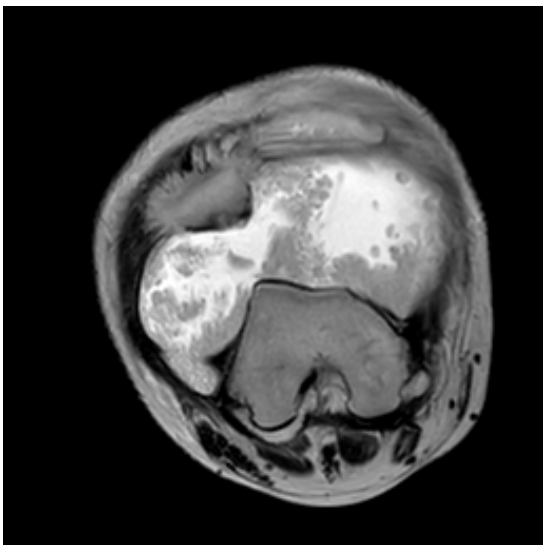

Figure 6: T2W TSE axial view showing synovial hypertrophy with villous and nodular protrusions and a large amount of fluid.

Figure 7: STIR longitudinal view showing synovial hypertrophy with villous and nodular protrusions and a large amount of fluid.

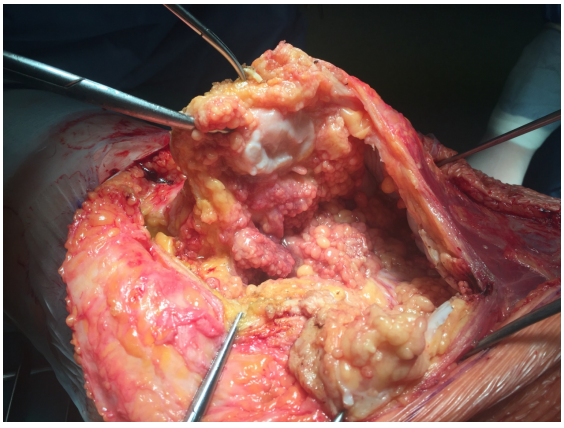

Figure 8: Pathologic examination of the removed synovial tissue depicts pigmented villonodular synovitis.

Discussion

The patient is a middle-aged male with all the major features of pacydermoperiostosis: pachyderma, peristalsis and digital clubbing. Joint involvement, (synovitis and effusion) mainly of the lower limbs, is very frequent in patients with pachydermoperiostosis and is associated with periostosis of the long bones, secondary osteoarthritis and possible dysregulations of endothelial activation [1,5,7]. The treatment for joint involvement is non- steroidal anti-inflammatory drugs, joint fluid aspiration and local glucocorticoid injection, and some cases synovectomy [1,4,5]. Arthralgia and arthritis are frequent in pachydermoperiostosis but painful synovitis, refractory to treatment, is rarely reported [1,7]. Our patient had important swelling of the right knee, refractory to non – steroidal anti-inflammatory drugs and repeated joint fluid aspiration with local glucocorticoid injection and finally he needed right knee synovectomy (15 years after left knee synovectomy). Pachydermoperiostosis is a very rare disease and also pigmented villonodular synovitis is a rare disease, so we wanted to report an unusual association of two rare diseases. The pathogenic mechanisms of this disease, pachydermoperiostosis and pigmented villonodular synovitis, are uncertain and probably different but we can speculate that the overproduction of growth factors as VEGF and PDGF, induced by the increased levels of PGE2 in patients with pachydermoperiostosis could have a pathogenic role also in pigmented villonodular synovitis.

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