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ORIGINAL STUDY

Spontaneous swelling of the ankle in a child : a difficult diagnosis

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The authors present a case of monoarticular localized pigmented villonodular synovitis (PVNS) in a 7-year old girl. PVNS is a rare benign disease of the synovial tissue. It is especially rare in ankles of children, with only 15 cases reported in literature.

The girl presented with swelling and pain in the left ankle since 4 weeks. The tentative diagnosis was made after a joint puncture and a MRI scan. A synovial mass with a brown-yellowish appearance was seen during the excisional biopsy. After removing the entire mass and without adjuvant therapy, no recurrence was detected after 12 months.

As it is very rare in children, PVNS is easily misdiagnosed. Early diagnosis is important to prevent bone and cartilage damage. A review of the clinical, radiological and therapeutical features of PVNS are presented.

Keywords : Pigmented villonodular synovitis ; pediatric ; monoarticular hydrops.

INTRODUCTION

Pigmented villonodular synovitis (PVNS) is a rare benign proliferative disease affecting the synovial tissue in the joint, tendon sheath, or bursa. The incidence is estimated to be 10/million and 4/million of respectively localized and diffuse PVNS *(16)*.

It typically affects people between 20-40 years old. The joint most commonly affected is the knee, in 66-80% of the cases, followed by the hip, ankle and shoulder. Symptoms are pain and swelling. Magnetic Resonance Imaging (MRI) is the current technique of choice for diagnosing PVNS *(2,8,21)*. Histology reveals synovial hypertrophy, multinuclear giant cells and macrophages loaded with haemosiderin and lipid. The haemosiderin deposition, a breakdown product of hemoglobin and the result of old hemorrhage, creates an important diagnostic key: it gives the typical brown-yellowish color macroscopically and it causes typical signal characteristics in MRI imaging *(11,20)*. The therapy of choice is a total resection and radiological follow-up to detect recurrence *(20)*. As PVNS is uncommon, it is generally not included in the differential diagnosis of the physician. Which, in return, can lead to a delay in treatment. PVNS is considered to be a benign lesion, but metalloproteinases expressed by PVNS can be locally aggressive for the periarticular bone and cartilage *(5,21)*.

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Therefore, a correct diagnosis is important. In this case report we present an atypical case of PVNS in a pediatric patient and we review the literature for similar cases.

CASE REPORT

A 7-year-old girl presented with a history of spontaneous swelling and pain in the left ankle since 4 weeks. She discovered the swelling because of one shoe being tight. No previous trauma had occurred. Her mother didn't notice any limping nor complaints of nightly pain. Clinically the ankle showed mild hydrops with no redness and no local warmth. Besides pain in deep plantar flexion, the active and passive range of motion was normal. The gait was normal. A previous performed ultrasonography showed hydrarthrosis with hypervascular synovial hypertrophy.

At first clinical presentation in the orthopaedic department, an ultrasound guided puncture and a MRI scan was performed. The aspirate was very viscous with a translucent-yellowish color which changed to a more hemorrhagic aspect at the end of the aspiration. Cell count was erythrocytes 206506 /µL, leucocytes 1280 /µL (93% mononuclear lymphocytes). The fluid obtained 8.80 mg/dl glucose and 2,500 mg/dl proteins. Gram stain, culture, and crystal analysis were negative.

The MRI (Fig. 1) scan showed a synovial mass (15mm x 13mm x 23mm) in the anterior recessus of the tibiotalar joint, hypo-intense on T1- an mild hyper-intense on T2- weighted images. It concerned a synovial hypertrophy with a nodular component. No clear haemosiderin deposits were seen.

Subsequently an incisional biopsy for an anatomopathological analysis was planned. While incising the tibiotalar joint, a solid soft tissue mass popped oud in one piece. Subsequently, the surgeon changed the strategy to an excisional biopsy (Fig. 2). The anatomopathological analysis confirmed our tentative diagnosis showing broadened synovia plain of mononuclear cells, multinuclear giant cells and haemosiderin loaded macrophages, leading to the diagnosis of localized intra-articular pigmented villonodular synovitis in a 7-year old ankle.

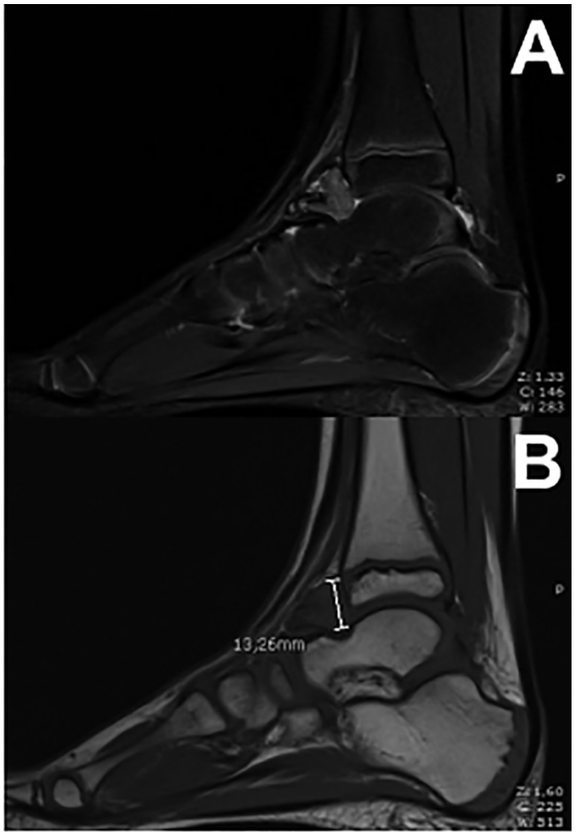

Figure 1. — MRI shows a well circumcised lesion in the anterior recessus of the tibiotalar joint. A. Sagittal fat-suppressed proton-density-weighted turbo spin-echo sequence with a hyper-intense signal of the lesion. B. Sagittal T1-weighted turbo spin-echo sequence with a hypo-intense signal of the lesion.

The patient was asymptomatic and had a normal ankle function 3 weeks after surgery. An MRI scan 3-, 6-, 12- and 24- months postoperative showed no signs of recurrence.

DISCUSSION

PVNS was first described by Chassignac in 1852 and the term PVNS was introduced in 1941 by Jaffe. In 1976 PVNS was subdivided by Granowitz into two forms : a diffuse form that affects the entire synovial membrane and a localized form that affects portions of a particular joint or tendon. The diffuse form is more prone to recurrence after surgical excision *(13,19)*.

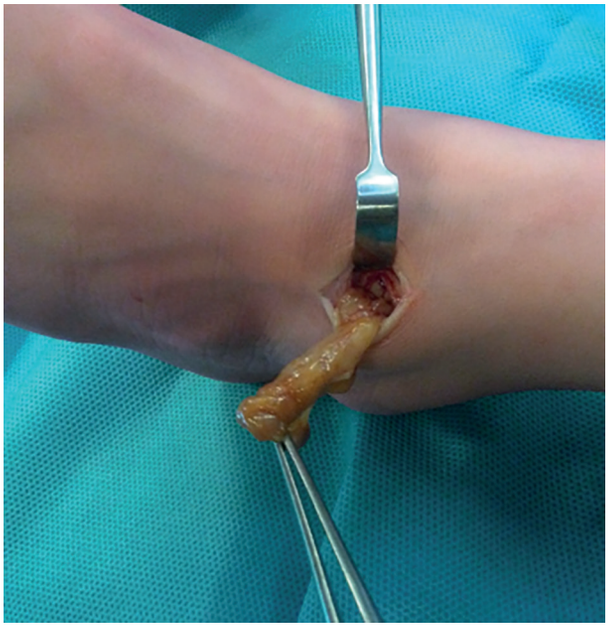

Figure 2. — Synovectomy of tibiotalar joint in a 7-years-old girl reveals a pigmented villonodular synovitis.

The etiopathogenic mechanism of PVNS remains a point of controversy and debate. Some authors suspect a neoplastic origin in the diffuse form because of monoclonality and chromosomal abnormalities *(5,21)*. Studies found PVNS cases with trisomy 7 *(10,23)*. This theory is also supported by reported cases of metastases *(9)*. There is also a lot of discussion about the genetic factors, especially in children, because of congenital anomalies or sibling involvement *(14)*. Another possible etiopathogenic mechanism is an inflammatory process, however the causative agent remains unclear *(14)*. It is suspected that localized PVNS is caused by a local granulomatous reaction, with again an unknown causative agent *(2,20)*.

PVNS is most frequently encountered in the third or fourth decade and is rare in children *(2,17)*. Approximately 2.5 to 20% of the cases of PVNS are located in the ankle, either in the joint synovium or in the tendon sheath *(4,17)*. To our knowledge, there are only 15 cases of unifocal PVNS of the ankle in children described in the literature (Table I). The reported cases consist of 4 boys and 11 girls whose ages ranged from 3 to 17 years at the time of presentation. All patients were treated with synovectomy. In one case the synovectomy was accomplished by arthroscopy, in the other cases an artrhotomie was performed. In most cases a local wide excision was performed but one case had an additional procedure involving tenosynovectomy of the tibialis posterior and the flexor digitorum longus tendon and one case underwent excision and curettage of the articular surfaces of the ankle. Only one patient had recurrence after 2 years. She was a 12 year old girl and the recurrence presented itself as an extra-articular mass of the synovial sheath of the fibular tendons. It required a surgical revision with synovectomy and a subtalar arthrodesis but a second recurrence appeared after 16 months. In most cases the time to diagnosis was 6 months to 2 years. One patient presented himself with acute pain and swelling and the diagnosis was made after three days. In another patient the diagnosis was made after 2 weeks. In our case the delay was only 4 weeks.

These cases show that there is frequently a delay in diagnosis of PVNS. The clinical symptomatology of PVNS is not specific and there are no pathognomonic signs, making the diagnosis difficult. The most typical symptoms are mono-articular swelling, sometimes palpable in the form of a mass, mechanical pain and loss of motion *(3)*.

Although very rare, the diagnosis should be considered in a child with an unexplained long-term swollen ankle. A joint aspiration can be helpful diagnosing the cause of a swollen ankle *(19)*. In this case the aspirate was aspecific (bloody and clear yellowish aspirate). Nonetheless, as such, we were able to rule out septic arthritis and some other causes like inflammatory arthritis became less likely. MRI is the imaging technique of choice. It shows a characteristic nodular synovial thickening and a typical aspect caused by the haemosiderin deposits. Other imaging techniques are aspecific. A radiography shows joint effusion and, in a later stage, bone erosions, but no typical signs. A diffuse hypo-echogenic nodule and thickened synovium can be seen on ultrasonography. A computed tomography arthrography can show some aspecific lesions and because of the radiation exposure, it is not the first choice *(20,21)*. Position emission tomography-computed tomography (PET-CT) is not traditionally used in the workup of PVNS. PVNS can be intense

Table I. – Reported pediatric cases of mono-articular pigmented villonodular synovitis in the ankle

Case	Author	Age	Gender	Joint	Time before diagnosis (years)	Symptoms	Treatment	Recurrence	Follow-up (years)
1	Bisbinas	15	F	L	2	Persistent swelling, minimal symptoms.	Arthrotomy. Local wide excision.	No	12.5
2	Bisbinas	15	F	R	2	Persistent swelling, associated discomfort.	Arthrotomy. Local wide excision.	No	2.5
3	Bisbinas	15	F	R	2	Persistent pain, less swelling	Arthrotomy. Local wide excision.	No	1
4	Bisbinas	12	F	R	1,5	Persistent swelling, mild pain.	Arthrotomy. Local wide excision.	No	2
5	Bisbinas	14	F	L	2	Persistent pain, less swelling.	Arthrotomy. Local wide excision.	No	1
6	Ganley	10	F	R	0,5	Persistent swelling, mild pain.	Local wide excision.	No	0,7
7	Aghasi	3	M	L	-	Abrupt onset of pain and joint effusion.	-	No	10
8	Kaneko	14	F	L	1	Pain and swelling.	Arthrotomy. Local wide excision.	No	3,3
9	Soifer	11	F	L	0,8	Swelling. No pain.	Arthrotomy. Local wide excision.	No	1
10	Neu-bauer	13	M	R	2	Pain and swelling.	Arthroscopy. Local wide excision.	No	2
11	Pannier	12	F	L	2	Pain and swelling.	Arthrotomy. Local wide excision.	Yes, after 2 years.	5
12	Sharma	8	F	R	2 weeks	Swelling. No pain.	Arthrotomy. Local wide excision.	No	2,7
13	Sharma	14	M	L	0,5	Swelling. No pain.	Arthrotomy. Local wide excision.	No	1,6
14	Sharma	17	M	R	3 days	Swelling, Acute painfully.	Arthrotomy. Local wide excision, synovectomy and curettage.	No. Awaiting triple arthrodesis for osteoarthritis.	2,8
15	Sharma	15	F	L	0,5	Swelling and pain.	Arthrotomy. Synovectomy of tibialis posterior and flexor digitorum longus tendon.	No	5,5

hypermetabolic on fluorine 18 fluorodeoxyglucose (18F-FDG) PET-CT but it's difficult to discriminate between malignant and benignant lesions. If an incidental intra-articular or juxta-articular lesion with atypical behavior is seen on 18F-FDG PET-CT, then there should be a low threshold for referral to MRI *(7)*.

In our case, MRI showed synovial hypertrophy but not the typical characteristics caused by the haemosiderin deposits, probably due to the early diagnosis.

The therapy of choice for PVNS is complete surgical excision, which can be performed by open or arthroscopic techniques. If it is a diffuse

PVNS, then a total synovectomy is recommended. Incomplete synovectomy is associated with high incidence of recurrence in such cases *(19)*. It is possible to use external beam radiotherapy and intra-articular radiocolloid injection to reduce the risk of recurrence, however this can result in a radiation-induced sarcoma). Recently new systemic therapies, such as colony stimulating factor 1 (CSF1) receptor inhibitors, have been proposed in the treatment of locally advanced or relapsed diffuse PVNS and as a preoperative or postoperative treatment of operable PVNS. Recent studies are promising but further investigations are necessary *(6)*. It is recommended to have clinical follow-up and MRI scans 3- and 12- months after surgery to detect recurrence *(15,21)*.

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