Pachydermodactyly may mimic juvenile idiopathic arthritis

Sir,

A 16-year old boy presented with a 3-year history of a painless, progressive, symmetrical swelling of the fingers, recently causing impairment while playing the guitar; there was a similar swelling on his chin since 6 months. General health was good. Family history was negative for similar changes or rheumatic disorders.

On clinical examination (Fig. 1) there were fusiform swellings limited to the ulnar and radial sides of the proximal interphalangeal joints and proximal phalanges of fingers II to IV and, less important of the radial side of finger V of both hands. The overlying skin was thickened, slightly erythematous and scaly. A similar swelling was present on the chin.

An ultrasound and X-rays of hands and chin showed soft tissue swelling with normal underlying bones and joints. Blood tests did not show inflammation nor the presence of antinuclear antibodies. A skin biopsy revealed hyperorthokeratosis, hypergranulosis and mild acanthosis of the epidermis. In the thickened reticular dermis there was an increase of irregular collagen bundles and mucine, but a loss of elastic fibres.

Based on these findings the diagnosis pachydermodactyly was made. After further questioning it became clear that the boy had a habit of crossing and rubbing his fingers followed by touching his chin when playing computer games. There was no evidence for an underlying psychiatric disorder. Three months after stopping this movement an obvious improvement was apparent (Fig. 2).

Pachydermodactyly is a benign superficial fibromatosis of the fingers, mainly affecting young males, first reported by Verbov (1) in 1975. About 60 cases are published until now, but it may be underreported.

The condition is characterized by symmetrical painless swellings over the radial and ulnar aspects of the proximal interphalangeal joints of the fingers II to IV, although atypical forms have been described.

Bardazzi *et al*. (2) proposed a classification with five different forms, including: classical pachydermodactyly, localised or monopachydermodactyly, pachydermodactyly transgrediens (extension to the palms or proximal fingers), familial pachydermodactyly, and pachydermodactyly associated with tuberous sclerosis.

The cause of pachydermodactyly remains unknown and may vary among patients. Repeated mechanical trauma (as in our patient) has been proposed in some children (2,3). As far as we know, regression after cessation of the mechanical trauma has been reported only once up till now (4). A compulsive neurotic personality (3) such as in Asperger syndrome (5) has been associated with pachydermodactyly, although this remains exceptional. Moreover pachydermodactyly seems exceptional in children with comparable compulsive disorders.

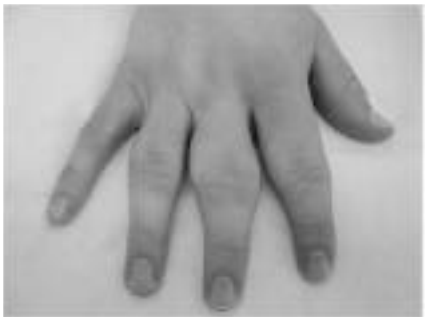

Fig. 1. Swelling of the fingers at the first visit.

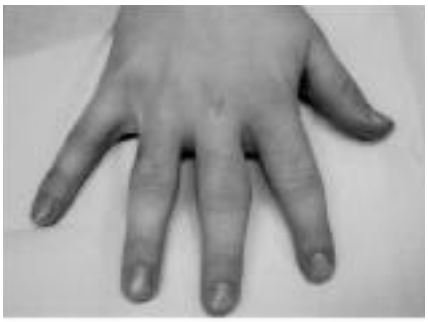

Fig. 2. Obvious improvement 3 months after stopping compulsive movements.

As an association with knuckle pads in the father has been described, it has been suggested that pachydermodactyly presents a variant of this disease, an underlying defect in proliferation of collagen being a predisposing factor (6).

Pachydermodactyly may be misdiagnosed as juvenile idiopathic arthritis (7). The absence of pain and morning stiffness, of synovitis on clinical examination, of inflammatory blood changes, together with the typical thickening of the skin are the most important differences with JIA. The differential diagnosis should also include true knuckle pads (6) which are fibromatous thickenings on the dorsum of the fingers joints; pachydermoperiostosis (8), a familial disease where bone and soft tissue proliferation produce clubbing and a spade-like enlargement of hands and feet, associated with thickening and furrowing of facial skin and juvenile digital fibromatosis, mostly occurring in infants as solitary or multiple soft tissue tumors at the digits of hands and feet.

No satisfactory treatment for pachydermodactyly has been described in the literature. Intralesional corticosteroids (9) and excision (10) are possible. Correction of a triggering compulsive habit may lead to remarkable improvement as illustrated by our case.

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Comparative analysis of hexosaminidase and cathepsin D expression in synovial fluid of patients with rheumatoid arthritis and traumatized joints

Sirs,

Recently, significant advances have been made in understanding the mechanism of articular cartilage destruction. Matrix metalloproteinases and cysteine proteases (cathepsins) are claimed to be the major enzymes implicated in this destructive process (1, 2). However growing data have been published on exoglycosidases as participants in the pathogenesis of joint damage (2-4). In the present paper we compare the activity of N-acetyl-β- hexosaminidase (HEX) and cathepsin D in the synovial fluid (SF) of patients with rheumatoid arthritis (RA) and juvenile idiopathic arthritis (JIA).

We examined patients with two types of rheumatoid diseases. 11 patients had RA (6 female, 5 male; 27-76 years old), with

Letters to the Editor

Table I. The activity of N-acetylo-β−hexosoaminidase and cathepsin D in synovial fluid of patients with rheumatoid arthritis (RA), idiopathic juvenile arthritis (JIA) and traumatized knees (p value means that the difference between the experimental and control groups was statistically significant.

Lysosomal enzymes	Enzymatic activity (mean) in the synovial fluid of patients with		
	RA n = 11	JIA n = 14	Traumatized knee (control group) n = 20
Hexosaminidase nM/ml/min	105.96 ± 34.04 p = 0.000003	85.13 ± 26.87 p = 0.0005	12.74 ± 4.25
Cathepsin D Tyr nM/ml/12h	146 ± 31.85 p = 0.000001	107.2 ± 21.19 p=0.00002	51.7 ± 16.42

swollen and painful knees during physical examination and effusion of the joint; duration of the disease was 4-28 years. The second group contained 14 patients with JIA(5 girls, 9 boys; 8-17 years old); six of them had polyarticular and eight oligoarticular onset JIA; duration of the disease was 1-11 years. At the time of sampling eight patients were in very active and six in subacute periods of the disease according to Mallya and Mace (5). Our reference group was composed of 20 patients with injured anterior ligament or meniscus medialis/ lateralis (6 female, 14 male; 15-21 years old).

Arthrocentesis of the knee joint of RA and JIApatients was performed because of prolonged exudation or intra-articular injection of steroids. Samples of the SF of patients with traumatized knees were obtained during routine diagnostic arthroscopy. The activity of HEX in the SF was determined as described by Zwierz *et al*. (6). The activity of cathepsin D in the SF was performed as described by Greczaniuk *et al*. (7).

Statistical analysis was conducted with a Statsoft program by Statistica 6, and the Levine test was applied to the data. This revealed significant differences among the studied groups. We used post hoc analysis calculated by test NIR, which indicated the least significant difference. Results were expressed as mean and SD. Pvalues of less than 0.05 were considered significant. The study design was approved by the Ethical Committee of the Medical University of Bialystok, Poland.

In the SF of patients with RAand JIA, HEX activity was calculated as 105.96± 34.04 nmol/ml/min and 85.13±26.87 nmol/ml/min respectively, and HEX activity in the RA group was significantly elevated (p = 0.000003) in comparison to traumatized patients (Table I). The above results are in agreement with our previously reported data (8).

The activity of cathepsin D in the SF of patient with RAand JIAamounted to 146.5 ± 31.85 Tyr nmol/ ml/ 12h and 107.2± 21.19 Tyr nmol/ml/12h respectively and in RA patients was significantly elevated (p = 0.000001) in comparison to traumatized patients.

Sohar *et al.* (9) recently reported a 1.28 fold increase in HEX, and 1.49 fold increase in cathepsin D activity in the leukocytes of RA patients. It is worthy of note that HEX activity in the SF of our RA and JIA patients was 8.3 and 6.6 but cathepsin D was only 2.8 and 2 times higher than the in the SF of the control group.

Our results suggest that in the knee joint cavities of RAand JIApatients, increased degradation of glycosaminoglycans (hyaluronic acid, chondroitin and keratan sulphates) and of glycoproteins with HEX is greater than degradation of proteins by cathepsin D. Our results are in agreement with data reported by Ortutay *et al*. (2) which suggest that exoglycosidases, which are present in the SF of RA patients, may contribute to the depletion of GAGs from cartilage. The conclusion is that some possible interplay between proteases and glycosidases in the SF of RA patients can take place. Analysis of the above enzyme system may be an important complement to molecular and genetic studies in the effort to fully understand the mechanism of RA. Our data indicate HEX as an important complement of the joint damage diagnostic system and inhibition of HEX as a potential target of RAtherapy.

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Atypical axial osteomalacia: Report of a HLA-B27 negative elderly female patient without features of sacroiliitis

Sirs,

Atypical axial osteomalacia (AAO) is a very rare bone disorder characterized by dense coarseness of the trabecular bone on radiographs located in the axial but not appendicular skeleton, and osteomalacia on bone biopsy of affected areas. Since the first case was described in 1961/62 (1) not more than 18 AAO-patients have been reported (2-9). All of them, except one (report of an affected mother and son) (5) were middle-aged or elderly Caucasian men. The cause of AAO is still unknown and some authors suggest that AAO could be a genetic bone cell abnormality. Sacroiliitis (3/5) and positive HLA-B27 antigen (2/4) is the most described concomitant disease. Axial increased and peripheral decreased bone mineral density (7, 9), moderate phosphate diabetes (8) and associations with polycystic kidney (5) and liver disease (5) have been reported.

A 83-years-old Caucasian female patient was admitted to our unit for dorsal and lumbar back pain. The patient was in normal general health, with no living relatives and no relevant medical or surgical history.

X-rays of the lumbar spine and pelvis showed a marked osteosclerosis without changes in size. There were no signs of sacroiliitis, which was confirmed by CAT, MR and scintigraphic examinations, whereby X-rays and MR are usually sufficient to make diag-