Articular manifestations of Scheie's syndrome

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Abstract

A patient with the clinical and biochemical features of Scheie's syndrome is reported. Radiological investigation has shown severe progressive juxta-articular cystic lesions, which have not, to our knowledge, previously been described in Scheie's syndrome.

Scheie originally described the syndrome attributed to him in 1962¹ as a variant of Hurler type mucopolysaccharidosis with excessive urinary excretion of dermatan sulphate and heparan sulphate as a result of a deficiency of α iduronidase. McKusick² and Watson-Jones³ reported carpal tunnel syndrome in this disorder and this paper describes another connective tissue abnormality.

Case report

A 60 year old retired musician was first seen at King's College Hospital in 1978 with impaired vision as a result of corneal infiltration and glaucoma. He had severe bilateral carpal tunnel syndromes with marked thenar wasting, which were treated by surgical decompression. His brother was also seen with identical symptoms and was treated in a similar way. A diagnosis of mucopolysaccharidosis type 1, or Scheie's syndrome, was made and this was confirmed by the marked deficiency of α iduronidase in his white blood cells (WBCs) and plasma (α iduronidase: patient WBCs 0.065, control 13.8

nmol/h/g protein), patient plasma undetectable, control 2.43 nmol/h/g protein).

During the ensuing years the patient's glaucoma was controlled by surgery on both eyes and with regular use of eye drops. He continued to play the piano, but his vision gradually deteriorated in his left eye. When seen in 1989 there was dense bilateral corneal opacification typical of mucopolysaccharide deposition.

On examination his corrected visual acuities were right 6/36 and left 6/60 and his reading vision was reduced to right N12 and left N18. He did not report having had any joint pain, but there was obvious bony swelling of many of the small joints of both hands. Extension of the wrist joints was limited and there were fixed flexion deformities of the terminal interphalangeal joints. Rheumatoid factor was negative and the concentration of uric acid in serum was normal. Radiographs of his hands and feet (figs 1 and 2) showed many obvious clear cut juxta-articular cystic lesions, some of which appeared to have fractured into the joints. He also had signs of a mild aortic stenosis, but there were no cardiac symptoms. An echocardiogram confirmed the presence of a calcified aortic valve with reduced opening.⁴

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Figure 1 Radiograph of left and right hands, clearly showing juxta-articular cystic lesions, some of which have fractured into the joints causing a destructive arthritis.

Figure 2 Radiograph of right foot, clearly showing juxta-articular cystic lesions, some of which have fractured into the joints causing a destructive arthritis.

His brother had died suddenly while walking to work at the age of 45 years and a necropsy showed aortic valve disease. Mild to severe narrowing of the coronary arteries can be found in patients with Hurler syndrome, so angiography was performed in May 1990. The results were normal.

Discussion

Scheie, an ophthalmologist, described early corneal clouding in ten patients and showed the deposition of acid mucopolysaccharide in corneal specimens.¹ The presenting problem of the patient reported here was corneal infiltration. As with Scheie's patients, he was of normal intelligence. He also had carpal tunnel syndrome, previously reported in patients with Scheie's syndrome associated with the excessive deposition of collagenous tissue around the median nerve.³ In addition, he showed the typical deformity of ankylosis of the distal interphalangeal joints of hands and feet and an echocardiogram confirmed mild aortic stenosis. What is unusual, and, to our knowledge, not previously reported in this disease, are the radiological findings in the hands and feet in a man with no other explanation for these changes such as gout or rheumatoid disease. It seems highly probable that there are deposits of mucopolysaccharide in his hands and feet, as described by Scheie in the cornea.¹ Previous

radiological investigations of patients with Scheie's syndrome have shown demineralisation and bony ankylosis of the distal interphalangeal joints. McKusick described a brother and sister with this disorder and commented on the cystic changes in a radiograph of the brother's hand.² These are by no means obvious in the published photograph. The only other previous report of severe bony changes is from Germany⁵ and the published photograph shows the complete absence of the carpal bones. This was not seen in our patient.

Conclusions

We suggest that this patient, with typical clinical and biochemical features of Scheie's syndrome, is the first to show severe progressive juxta-articular cystic lesions in the hands and feet. In the absence of any other disease process we attribute this to Scheie's syndrome.

- 1 Scheie H G, Hambrick G W, Barnes L A. A newly recognised forme fruste of Hurler's disease (Gargoylism). *Am Ophthalmol* 1962; **53**: 753-69.
- 2 McKusick V A. *Heritable disorders of connective tissue*. 4th ed. St Louis: Mosby, 1972: 548-56.
- 3 Watson-Jones R. Leriche pleonosteosis, carpal tunnel compression of the median nerves and Morton's metatarsalgia. *J Bone Joint Surg* 1949; **31**-8: 560-71.
- 4 Schinz H R, Furtwängler A. Zur Kenntnis einer hereditären Osteoarthropathie mit rezessiven Erbgang. *Deutsche Zeitschrift Chirurgie* 1928; **207**: 398-416.
- 5 Leroy J G, Crocker A C. Clinical definition of the Hurler-Hunter phenotypes. A review of 50 patients. *Am J Dis Child* 1966; **112**: 518-30.