Sex-influenced expression of Madelung’s deformity in a family with dyschondrosteosis

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SUMMARY  Dyschondrosteosis is a mesomelic form of short stature which occurs in conjunction with a characteristic wrist deformity, Madelung’s deformity. A family with dyschondrosteosis had an affected father and two daughters. The affected females had dyschondrosteosis and Madelung’s deformity, while the affected males had dyschondrosteosis, but no Madelung’s deformity. All affected members had arthralgias. The occurrence of male to male transmission confirms an autosomal dominant inheritance pattern for this disorder.

Dyschondrosteosis is a skeletal dysplasia consisting of Madelung’s deformity and mesomelic dwarfism, with little or no involvement of the vertebrae.1 Previous reports have noted a 4:1 female predominance and that females were more severely affected than males. Since no documented cases of male to male transmission have been observed, this has been classified as having either an autosomal dominant inheritance or an X-linked dominant inheritance. Few families have been ascertained in the United States.

In this report we present a family with dyschondrosteosis inherited in an autosomal dominant pattern, but in which Madelung’s deformity was present only in the affected female members. All of the affected members had arthralgias in involved joints indicating that this disease, like many other skeletal dysplasias, predisposes to degenerative joint disease.

Case reports

The pedigree of the K family is presented in fig 1.

DK (I.1), aged 40, had been in good health throughout his life, but had always noted an inability to extend his wrists and tightness in his hands. He had been told he had Osgood-Schlatter disease as a child and had a 10-year history of arthralgia in his left knee. He was a physical education teacher and was able to participate in most sports.

On physical examination, his height was 167 cm. He had mesomelia of the arms and legs and could not extend at either wrist past 45°. He had stiffness in all joints. There was crepitance and pain on motion of his left knee. He had prominence of the medial tibial tuberosity. The remainder of the examination was normal.

KK (I.2), the mother aged 35, had been in good health all her life. She was 156 cm tall, but was normally proportioned and physical examination was normal.

AK (II.1), the eldest sister aged 22, had been noted since birth to have deformed wrists. She was always short but otherwise in good health. She had been diagnosed as having Osgood-Schlatter disease as a child. On physical examination, she was 147 cm tall, has mesomelia, and Madelung’s deformity in both wrists, with the left wrist more severely involved than the right (fig 2a). She had prominent medial tibial tuberosities.

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Jack R Lichtenstein, Murali Sundaram, and Robert Burdge

FIG 2 The left wrists from (a) II.1 and (c) II.4, sisters, who have dyschondrosteosis and Madelung's deformity, and (b) II.2, brother, who has dyschondrosteosis but no Madelung's deformity.

HK (II.2), the eldest brother aged 20, had always been small, but otherwise in good health. He had had low back pain for several years. On physical examination his height was 160 cm and he had mesomelia, but did not have Madelung's deformity (fig 2b).

CK (II.3), aged 19, was an unaffected brother who was in good health.

GK (II.4), the proband, a 12-year-old female, was ascertained because of limitation of ability to do certain exercises such as push-ups and wrist pain. She had otherwise been in good health except for multiple dental extractions because of crowding. On physical examination her height was 130 cm. She had Madelung's deformity bilaterally (fig 2c) and mesomelic dwarfism. Examination was otherwise normal.

The family history revealed no other affected members. The father had been adopted and was unaware of other members of his family. There was no evidence for ocular, auditory, or dermatological difficulties in any of the affected members.

X-RAYS

X-rays of II.1 and II.4, the affected sisters, showed the following characteristics of Madelung's deformity (fig 3).3 4

1. A shortened radius in relation to the ulna.
2. Dorsal subluxation of the ulna.
3. Triangularisation of the distal radial epiphysis.

FIG 3 (a) and (b) x-rays of the forearms of II.1 and II.4 showing Madelung's deformity, (c) x-ray of the forearm of II.2 showing absence of Madelung's deformity.
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(4) An ulnar and ventral slant of the radial articular surface.
(5) Triangularisation of the carpal bones.

The affected sisters also had deformed medial tibial plateaux with exostosis at the margin. The two affected males with mesomelic short stature did not have radiographic evidence of Madelung’s deformity. I.1 had typical changes of osteoarthritis of both knees. X-rays of the mother (I.2) were normal.

Discussion

Dyschondrosteosis is a rare skeletal disease characterised by mesomelic dwarfism and Madelung’s deformity. Twelve criteria have been detailed for diagnosing Madelung’s deformity, with the primary criteria being foreshortening of the radius, lucency of the distal radial epiphysis, triangulation of the carpal bones, and posterolateral displacement of the radial stylos. Exostoses have also been noted on occasion. Most of the 12 criteria are radiographic; however, the anomaly can be identified clinically by the posterior displacement of the distal ulna and limitation of flexion and extension at the wrist.

In this family mesomelic dwarfism was inherited by autosomal dominant transmission. The transmission from father to son indicates definite autosomal dominant inheritance. In addition, the two affected females had Madelung’s deformity, while the two affected males did not. A previous report has noted a family in which an affected male member had involved mesomelia without Madelung’s deformity, while two females had both Madelung’s deformity and mesomelic dwarfism. They suggested that this disorder has less severe manifestations in males than in females. In our family, one of the affected males (II.2) had more severe mesomelic dwarfism than either of the affected females, but did not have Madelung’s deformity. His father (I.1) had mild mesomelia without Madelung’s deformity.

In contrast, both of the affected female members had prominent Madelung’s deformity as well as mesomelia. Since Madelung’s deformity is a congenital anomaly, this suggests that its development is influenced by in utero sex hormones or by X chromosomal genes.

A second interesting feature is the prevalence of significant arthralgias in affected family members; two members had been diagnosed as having Osgood-Schlatter disease. All of the affected members complained of wrist pain, and one had significant low back pain. Dyschondrosteosis should be included among the group of skeletal dysplasias which cause premature osteoarthritis.

References


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