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一个Schmid型干骺端软骨发育不良症家族

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摘要 本文报道了在安徽省宿县地区发现的一个Schmid型干骺端软骨发育不良症家族。调查涉及五代，患者10人（6男，4女），现存活6例（4男，2女），经家谱分析，符合常染色体显性遗传。患者智力、外貌、头颅、脊椎均正常，但四肢短小，指（趾）粗短，弓形腿，髋内翻，步态摇摆。男性患者的腕、踝等关节有明显畸型；而在这些部位，女性患者无明显畸型，其它症状也较男性患者为轻，说明男女患者的表现度明显不同。

关键词 Schmid型干骺端软骨发育不良,弓形腿,髋内翻,短肢侏儒

分类号

A Kindred of Metaphyseal Chondrodysplasia, Schmid Type

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Abstract

This paper reports a kindred of metaphyseal chondrodysplasia, Schmid type, which has been found in an area of north Anhui. Five generations in the kindred have been investigated, in which six (4 males and 2 females) out of the ten affected individuals (6 males and 4 females) are stiu living. The pedigree presents autosomal dominant inheritance. The patients are short-limbed dwarfism with normal intelligence, head, looks and spina, but bowlegs and coxa vara. Irregularities of the metaphyseal ends of bones in the extremities can be demonstrated by X-ray examination. Carpal bone and anklebone show obvious edformation in the male patients besides short limbs. The limb deformation and other symptoms are, however, more severe in affected males than those in affected females. It indicates that there exists difference between the expressivities of males and females.

Key words Metaphyseal chondrysplasia Schmid type Bowlegs Coxa vara Short-limbed dwarfism

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