LETTERS TO THE EDITOR

Male proband with X linked retinoschisis apparently inherited from his father's family

We would like to draw attention to two points in relation to a family with X linked retinoschisis. First, an affected male with an unaffected father but an affected maternal uncle should not be concluded to have inherited his otherwise consistently X linked condition by some anomalous mechanism from the male side of his family: the possibility that his mother is a carrier should be immediately considered. Second, the public records, in this case in Register House, Edinburgh, Scotland, can be valuable in establishing the characteristics of the pedigree and hence in supporting the diagnosis.

Juvenile X linked retinoschisis is an uncommon disease of the retina which derives its name from the splitting of the retina very superficially in the nerve fibre layer so that a very thin veneer of retina bulges forwards. Fenestrations in this thin retina are frequent and may cause retinal blood vessels to rupture, hence vitreous haemorrhage. In 50% of cases the macular area is involved, which reduces the visual acuity. There are multiple radiating lines of superficial cystoid spaces around the fovea early in the disease, but these later coalesce to produce a larger cavity and eventually, in the elderly, a thin atrophic macula.^{1,2} A new textbook contains colour pictures.³

Our pedigree belongs to the same family as originally described by Levy⁴ and also briefly mentioned by Collyer and de Mey.⁵ A recent study of four Swedish families with juvenile retinoschisis (RS) locates the abnormal gene at Xp22 and suggests the gene order for the series of closely linked genes to be (OTC the most proximal): *DXS85*–(*DXS43*, *RS*, *DXS41*)–*DMD*–*DXS84*–*OTC*.⁶

During a diagnostic survey of the Royal Blind School, Edinburgh,⁷ we noted three brothers with this disease, inherited from a carrier mother with maternal relatives affected as in X linked disease (fig 1). A curious piece of information came to light, in that the

carrier mother's oldest brother (unaffected) had a blind son. Although we assumed that the cause of this blindness could not have been X linked retinoschisis, we decided to examine him. To our surprise, he too had retinoschisis which he seems to have inherited through an unaffected father, an inexplicable phenomenon.

His mother did not know as much about her family as her husband did about his family. However, she believed that her late father had been blind. She also said that someone had once told her that she was a distant blood relative of her husband. Accord-

ingly, the records of Births, Marriages and Deaths for Scotland at Register House were scrutinised at the General Register Office, New Register House, Edinburgh.

The families were traced back for six generations and both families (paternal and maternal) were found to originate from one ancestral couple born c1819 and c1803 (fig 2). A daughter of this couple was the paternal great great grandmother (born c1843) and a son the maternal great great grandfather (born c1854) of the proband. Statutory compulsory registration of births was not introduced until 1855,

Figure 1 The proband, born in 1963, was blind from the same disease, retinoschisis, as his three cousins. This X linked recessive disease appears to have been inherited through an unaffected father. See fig 2 for the explanation.

Figure 2 Patient VII.1 has inherited his abnormal X chromosome from his mother who has a degree of consanguinity with his father.

but on examination of the marriage certificates of these two sibs the names of their parents were found to be identical, and their relationship was confirmed from the 1861 census when they were living in the same household and were recorded as brother and sister. Unfortunately, we have no documentary evidence that the brother was affected. Their mother (I.1, fig 2) was born in 1819 and can be quite confidently presumed to be a carrier whose abnormal X chromosome was inherited by these two children. Accordingly, the proband had not inherited his disease from his normal father's side of the family but from his carrier mother. This case shows the importance of taking a full family history of both sides of the family and of enquiring about consanguinity. We might have missed this diagnosis if the maternal grandmother had also been a carrier (with a normal, instead of an affected, husband), a possibility often forgotten in relation to X linked disease. Conversely, without the details available from Register House, we might have postulated some highly speculative mechanism whereby the proband inherited his X linked disease through an unaffected father.

In summary, we were greatly exercised to try to find an explanation for X linked retinosis in a male proband with a typical X linked pedigree on the paternal side, especially as his own father was unaffected (though a paternal uncle was blind). The solution to the problem was achieved by gentle persistence in history taking from the mother, who believed that her father who died when she was very young was blind, and that she had once been told that she was a distant relative of her husband. Tracing of both families in the public records system of Scotland established that the proband's paternal great great grandmother was a sister of the maternal great great grandfather, with X linked recessive blindness in descendants of both.

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CHILD naevus is not ILVEN

In a recent article, Moss and Burn¹ advanced the hypothesis that CHILD syndrome and ILVEN are "polar groups on a clinical spectrum, both reflecting an ectodermal defect variable in site and extent". They proposed the new descriptive term 'psoriasiform epidermal naevus (PEN)', sometimes associated with 'congenital ipsilateral limb defects (PENCIL)'.

For the following reasons, however, the equation 'CHILD naevus + ILVEN = PEN or PENCIL' is mistaken. The epidermal naevus associated with the CHILD syndrome is definitely not ILVEN but a distinct cutaneous entity that should be called 'CHILD naevus'.²

(1) CHILD naevus can be distinguished from ILVEN by the presence of yellow, wax-like scales, resulting in a distinctive 'ichthyosiform' appearance. (2) This naevus shows a tendency to non-linear arrangement, often involving one half of the trunk in a diffuse manner. By contrast, ILVEN is always linear. (3) This naevus displays a pronounced affinity for the body folds, or ptychotropism³; by contrast, ILVEN is not ptychotropic. (4) CHILD naevus causes no, or only minimal, pruritus whereas in patients affected with ILVEN itching often constitutes a serious problem. (5) CHILD naevus may show the histopathological features of 'verruciform xanthoma', a phenomenon characterised by abundant foamy histiocytes occupying the dermal papillae. Such

xanthomatous transformation has so far not been observed in ILVEN. (6) CHILD naevus is a well defined genetic entity inherited as an X linked dominant trait, constituting the cutaneous hallmark of the CHILD syndrome. It occurs almost exclusively in females because the underlying X linked mutation is lethal in male embryos. By contrast, the genetic basis of ILVEN is unclear and possibly heterogeneous.

Furthermore, I disagree with Drs Moss and Burn that CHILD should apply only to the extreme 'hemidysplastic' form of a variable condition. It is true that the term was originally suggested as an acronym for 'congenital hemidysplasia with ichthyosiform erythroderma and limb defects',³ but it has now become evident that the associated skin disease should be classified more appropriately as a naevus, and therefore the following modified interpretation of the acronym has been proposed: 'congenital hemidysplasia with ichthyosiform naevus and limb defects'.⁴

In conclusion, there are different epidermal naevi giving the impression of a psoriasiform skin lesion, and it seems unjustifiable to lump them together under the term 'psoriasiform epidermal naevus'.

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The variable clinical spectrum and mental prognosis of the acrocallosal syndrome

In the August 1990 issue of this journal we read with interest the paper 'How wide is the clinical spectrum of the acrocallosal syndrome? Report of a