Discussion

Lymphoedema, when inherited, develops due to the action of a dominant autosomal gene[5] with incomplete penetrance of about 50%. There is sex influence with a higher proportion of females developing lymphoedema (M:F=0·4). In the small number of patients with bilateral hyperplasia the sex influence seems to be reversed, with males at greater risk than females (M:F=1·7).

The possibilities that could explain the occurrence of distichiasis with bilateral hyperplasia are (1) that there is linkage of two genes, (2) that there are two pleiotropic effects of the same incompletely penetrant gene with variable expression, or (3) that this is simply association.

Distichiasis is a rare condition and it does occur separately from lymphoedema. Fox surveyed published reports in 1962[6] and found 78 cases described. He found a strong hereditary influence without any sex predeliction but did not mention any connection with lymphoedema. McKusick[7] quotes Maumenee (personal communication, 1982) that it is only where lymphoedema is also present that distichiasis is inherited. Published reports are confusing and this may be due to the variability of expression of the two traits, particularly the late onset of lymphoedema in many cases.

Bilateral hyperplasia is a rare form of a rare condition and it is unlikely that distichiasis would only appear in this small group from the effect of linkage. There is no reason to think that the combination has any effect on survival and linkage disequilibrium cannot explain the findings.

Association is an unlikely explanation for the two conditions to appear together as they are both rare and one would have expected distichiasis to occur with other forms of lymphoedema.

Conclusion

The appearance of distichiasis with primary lymphoedema is restricted to those with bilateral hyperplasia of the lymphatics. If distichiasis is seen in someone with lymphoedema, it is likely that patient has bilateral hyperplasia rather than other forms of lymphoedema. Distichiasis and bilateral hyperplasia appear to be inherited through the action of an incompletely penetrant gene with two pleiotropic effects and variable expression.

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Correction

In the paper 'Carrier detection and prenatal diagnosis in X linked muscular dystrophy using restriction fragment length polymorphisms' by Lindlöf *et al,* published in the December 1986 issue of the Journal (*J Med Genet* 1986;**23**:560–72), an error occurred in fig 1. The pERT87 alleles underneath III.1 in family 5 should read S2, F2 and not S1, F2.