Ectodermal dysplasia in females

SIR,

I would like to comment on some points in the introduction of the recent paper by Moreno Fuenmayor *et al.*¹ It does not seem correct to say that "several variant forms" of the so-called "hypohidrotic ectodermal dysplasia" have been described and are "characterised by the presence of associated findings and different modes of inheritance". They are *not* "variant forms", they are different conditions, both clinically and genetically. The use of the expression "variant form" is misleading in this context and should only be employed when referring to different clinical "forms" of the same condition. This is a simple and important notion in nosology.²

Secondly, the name given to the condition "hypohidrotic ectodermal dysplasia" is also misleading, since there are a number of conditions that could be given this label,^{3,4} of which our recent review⁵ lists about 40. Therefore, a specific name should be given to each one of them to avoid confusion. For this reason, we prefer the old and well known eponymic designation, Christ-Siemens-Touraine (CST) syndrome.^{6,7} As a matter of fact, this condition is more than a pure dysplasia; it is a complex ectodermal dysplasia/malformation syndrome.

It is not true that "occasional minor manifestations" may be seen among carriers. As many as about 70% of carriers may be recognised^{7,8} and this is an excellent guide for genetic counselling.

Severe manifestation of CST-like conditions has been reported in females. There are three possible explanations for the existence of these women: heterozygous manifestation of the X linked syndrome owing to skewed X chromosome inactivation, homozygosity for the same X linked gene, and homozygosity for the gene of the clinically indistinguishable autosomal recessive ectodermal dysplasia.^{9,10} We described two severely affected females with CST⁷ and suggested a skewed inactivation of the X chromosome carrying the normal gene to explain these findings.

There is an equal probability of finding "normal" as well as severely affected carriers.

Regarding the three apparently normal carriers shown in the pedigree, the authors state that "dentition seemed normal but a complete dental history could not be obtained". The teeth are generally affected among carriers and therefore this

statement makes it less probable that they really are normal.

Finally, we would like to call attention to a spelling mistake: the correct words are anhidrotic and hypohidrotic (*not* hypohydrotic). Hydor means water and hidros means sweat.¹¹ That is why we write hydrogen, hydrocephalus, hydrolysis, etc, but hypohidrosis, hidrorrhoea, hidradenoma, etc.

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Delineation of trisomy 9

SIR,

The report in the October 1981 issue of *Journal of Medical Genetics* (18: 377-82) by Mantagos *et al* has added additional information to the delineation of trisomy 9. Numerous reports have clearly delineated