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

the 9p trisomy, 9q trisomy, and 9p monosomy conditions. However, few cases have been described to delineate the trisomy 9 syndrome. In fact, non-mosaic trisomy 9 has rarely been reported in term infants¹⁻³ and only once by prenatal diagnosis.⁴ I would like to elucidate further the anatomical dysmorphology and variability of this syndrome by reporting the prenatal diagnosis and subsequent anatomopathological examination of a trisomy 9 fetus.

Although advanced parental age has not been a hallmark of this syndrome, the ages of the mother and father of our case were 45 years and 41 years, respectively. There was no history of consanguinity and no family history of birth defects, mental retardation, or multiple miscarriages. Elective termination of pregnancy was accomplished at 19 weeks' gestation.

The male fetus delivered was grossly abnormal, weighing 176 g, with a crown/heel length of 19.5 cm, a crown/rump length of 13.9 cm, and a head circumference of 14 cm.

The facial appearance was characterised by a high forehead with flat supraorbital ridges, hypertelorism, enophthalmus, and bilateral corneal opacities. The nasal bridge was broad, the tip bulbous, and the nares anteverted. There was a poorly demarcated philtrum and cupid's bow, and marked micrognathia. The pinnae were small, simple, and low set. The neck was short and broad with mild webbing.

There was complete absence of the left hemidiaphragm. The heart had a double outlet right ventricle with a small but normally formed aortic valve, normal aortic and pulmonary outflow tracts, and a membranous ventricular septal defect. The mediastinum was shifted to the right and the hypoplastic left lung was approximately one-third the size of the right lung. The left hemithorax was occupied by the liver, spleen, stomach, and segments of the intestine. Apart from the retroperitoneal viscera only the descending colon and the greater portion of the liver remained in the abdomen. There was a small closed lumbar meningocele of 1 × 1 × 0.2 cm (amniotic fluid α -fetoprotein was reported to be within normal limits). There was complete lack of gyration of the cerebral hemispheres and absence of the optic tracts.

Although the facial appearance of the fetus reported here bears striking resemblance to those previously reported, corneal opacities, meningocele, and diaphragmatic hernia have not been previously noted. Though the eyes were not fully examined, the absence of optic tracts coupled with the appearance of enophthalmus suggest microphthalmia. Cardiac anomalies have been a frequent finding, including another case of double outlet right ventricle. Cystic

kidneys, a finding in other cases, were not seen in this case.

I wish to stress the importance of reporting cases of rare autosomal aneuploidy, for they help to facilitate meaningful genetic counselling.

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Wishes of patients with retinitis pigmentosa concerning genetic counselling

SIR,

It was first established by Emery and Smith¹ that only a minority of patients at high risk for transmitting a serious disease actually receive genetic counselling. At that time it was not known whether the remainder wished to have genetic counselling or not, but since then experience with registers for Duchenne muscular dystrophy and Huntington's chorea,^{2,3} and experience with a genetic service for handicapped school leavers,⁴ has shown that the majority of such patients or their families welcome the offer of genetic counselling.

For the last four years, patients with retinitis pigmentosa have been visited as part of a study to determine its incidence and heterogeneity within the City of Birmingham.⁵ The opportunity was taken to ask every index patient his or her views on genetic counselling. Some patients were severely affected with loss of vision in their thirties and forties, but the majority of patients (those with dominant or non-genetic retinitis pigmentosa) had a mild disease with preservation of useful sight in their fifties and sixties.

There were 142 index patients who were visited, although more patients are in the incidence study in which secondary cases are included. Twelve patients did not give their views on genetic counselling as they considered them to be irrelevant: four because they were unmarried and eight because they had