the proband did not have any cardiac anomaly, mental deficiency, or deafness.

Another point we want to make is that although our common abnormalities were similar to the cases of Glanz and Fraser, especially malformations of the toes, others, such as spina bifida, vitiligo, hypoplastic spleen, and scoliosis, were not noted.

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References

This letter was shown to Drs Glanz and Fraser who reply as follows:

We thank Drs Akar and Gözdaşoğlu for drawing attention to their paper and apologise for its omission from our review. We included café-au-lait spots as hyperpigmentation.

Philtrum length and intercommissural distance in newborn infants

Sir,

In their very useful paper, Sivan et al1 have provided normal values for philtrum length and intercommissural distance in newborn infants. I am glad to know that their standards correspond well with those I previously found in healthy Hungarian neonates.2 However, since gestational age cannot always be determined and intrauterine growth retardation is characteristic of many congenital syndromes and malformations, the consideration of gestational age alone may be misleading in a significant proportion of pathological infants.

Therefore, I would recommend that gestational age and birth weight should be taken into account. This has been our policy in determining standards for several measurable minor malformations and variants so far.

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References

Genetic heterogeneity in Duchenne muscular dystrophy

Sir,

From the results of linkage studies using two restriction fragment length polymorphisms, Dr O’Brien and colleagues1 question our earlier suggestion that Duchenne muscular dystrophy (DMD) may exhibit genetic heterogeneity.2 We would disagree with the interpretation of their results for several reasons.

Firstly, our suggestion was that a subgroup of DMD may possibly exist in which affected boys have severe (which we stressed) mental handicap usually requiring institutional care. Such boys in fact represent only a very small proportion of all affected boys. Over the years we have collected information on 302 affected boys (of whom 182 have been personally examined), but fewer than 20 have been classified as severely mentally handicapped. In Dr O’Brien’s study it is not clear in how many kindreds the affected boys were severely mentally handicapped, and in several kindreds data from mentally handicapped boys and boys with normal IQ were combined, which would not seem justified.

Secondly, as was discussed in our paper, there is already some convincing biochemical evidence of heterogeneity in DMD, though it is not clear if this is related to the observed clinical differences.

Thirdly, as the authors themselves point out, linkage studies cannot exclude heterogeneity resulting from different mutations at the same or closely linked loci. From what we know already about the fine structure of disease loci, if genetic heterogeneity in DMD does exist it would seem quite reasonable that it could be within a single locus.

For these various reasons we feel that the case against heterogeneity in DMD is therefore still ‘non-proven’. The situation is likely to remain unclear until such time as the molecular defect(s) has been characterised.

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References