Deletion Xp22.3

Dr Jean Frézal has kindly drawn our attention to an abstract by Friedman *et al*¹ describing a female infant heterozygous for a deletion involving band Xp22.3 who had features in common with the patients we described in the January issue of the Journal^{2,3} (microphthalmia and skin defects of the head and neck). In the patient of Friedman *et al*¹ the skin defects were said to extend onto the extremities, growth was normal (it was reduced in two of our three patients), and there was agenesis of the corpus callosum (not looked for in our patients). We feel certain that this is an earlier report of the syndrome that we described, but we are still not convinced that it is Goltz syndrome, as Friedmann *et al* suggest.

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- 1 Friedman PA, Rao KW, Teplin SW, Aylsworth AS. Provisional deletion mapping of the focal dermal hypoplasia (FDH) gene to Xp22.31. *Am J Hum Genet* 1988;43:50A.
- 2 Temple IK, Hurst JA, Hing S, Butler L, Baraitser M. De novo deletion of Xp22.2→pter in a female with linear skin lesions of the face and neck, microphthalmia, and anterior chamber eye anomalies. *J Med Genet* 1990;27:56-8.
- 3 Al Gazali LI, Mueller RF, Caine A, *et al*. Two 46,XX,t(X;Y) females with linear skin defects and congenital microphthalmia: a new syndrome at Xp22.3. *J Med Genet* 1990;27:59-63.

Families with X linked liver glycogenosis owing to phosphorylase kinase deficiency

Linkage analysis of X linked liver glycogenosis (XLG) owing to phosphorylase kinase deficiency (McKusick 30600) suggests linkage of the XLG gene to a cluster of marker loci on the short arm of the X chromosome (unpublished data). To exclude linkage heterogeneity we would like to study more families affected with this disease. If any reader has a family affected with XLG, we would like the opportunity to perform linkage analy-

sis. If you would be willing to cooperate please contact me at the address below.

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Medical genetics in Romania

Through a genuine revolution Romania has succeeded in getting free from a dictatorial system that, among many other crimes, hindered the development of medical education and research.

Human and medical genetics, which has made remarkable progress recently, is one of the fields in which nothing has been done in Romania in the last 15 years. We can never forget that we were not allowed to talk about Mendel and Morgan even before 1965.

The few Romanian specialists, trained in the 1960s and 1970s in various genetic centres in Europe and the USA, could not turn their knowledge to account. We had no foreign currency to obtain information, apparatus, reagents, etc. We did not have the right or the means to go abroad, to take part in scientific meetings. Our correspondence was censored, and we were not permitted to be visited by our foreign colleagues. To train young researchers or specialised physicians in medical genetics was a utopian idea, and any attempt in this direction was either ignored or punished. Some people preferred to leave the country in order to use their knowledge. We stayed here trying to do something and anticipating the day that finally came. However, we now start freely and hopefully a period of reconstructing education and research. It is difficult and we badly need any kind of friendly help.

At present, in Romania, there are medical genetics centres in Bucharest, Iași, Cluj, Timișoara, Oradea, Craiova, and Tîrgul Mureș. There are offices for registration and diagnosis of congenital anomalies in all the districts of our country. The number of specialists, information, apparatus, reagents, and funds are very reduced. Prenatal diagnosis is almost non-existent; as for gene diagnosis, our knowledge is limited to the very few things taken from the few books and papers we have received owing to the kindness of some colleagues.

There have been no subscriptions for journals of human and medical genetics either in Bucharest or elsewhere in this country.

We are writing these lines in grief and sadness hoping that you will understand our situation and our desire to bridge the gaps, with the aim of developing medical genetics. Appealing to the solidarity of the scientific world we ask your support for the following.

(1) The achievement of a plan and logistical programme for developing centres of medical genetics by offering us your opinions and your advice on organisation and priorities.

(2) Scientific documentation: books, monographs, and genetics journals (medical and especially human).

(3) Ways of improving academic education concerning genetics (manuals, video apparatus and cassettes, film slides on genetics) and ways of training young postgraduate researchers and specialists.

(4) Technologies of genetic research at the cellular and molecular level, including prenatal diagnosis, the necessary apparatus, and reagents.

(5) Computer programs in the field of diagnosis of malformation syndromes.

We are asking you kindly to express your point of view on each of the five problems and to help us, as well as to pass on our request to other institutions. Further information can be obtained from the following addresses.

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