LETTERS TO THE EDITOR

Kuwait type faciodigitogenital syndrome

In 1988 we¹ reported a newly recognised autosomal recessive malformation syndrome with some resemblance to Aarskog type faciodigitogenital syndrome, including short stature, hypertelorism, short, stubby nose with anteverted nostrils, long philtrum, ear anomalies, long neck with sloping shoulders, small, broad hands with mild interdigital webbing, fifth finger clinodactyly, lax hand joints, and shawl scrotum. However, the face was somewhat triangular or elongated, the eyes did not slant downwards, and there was no ptosis. In addition, the hair was coarse, dry, and hypopigmented in four of the five affected sibs (three male and two female).

In the large Kuwaiti Bedouin tribe (approximately 10 000) from which this family comes, after the publica-

tion of our paper, we observed another sibship with two affected males who are double first cousins to the reported cases (figure). Moreover, we also observed three other distantly related sibships with nine affected subjects who all can be traced to a common ancestor. This raises the total number of cases encountered so far to 16, including nine males and seven females, with ages ranging from 6 months to 10 years. This reflects a very high prevalence for a monogenic malformation syndrome in this tribe (at least 1 in 625) and if age specific minimum prevalence is considered it would be even higher. It is also noteworthy that these cases all live in Farwania district with a population of 400 000 only 15% of which is Bedouin. If the minimum prevalence is estimated among the Bedouin community in that district it would be 1 in 3750 (16:60 000) which is four times the frequency of Bardet-Biedl syndrome among the Bedouin.²

The consistent manifestations are the triangular or elongated face, telecanthus or mild hypertelorism, wide palpebral fissures without ptosis, short, stubby nose with anteverted nostrils, high arched and narrow

palate, long, deep philtrum, wide mouth with protruding lower lip, posteriorly rotated ears with minor ear anomalies, long neck with sloping shoulders, small, broad hands with mild interdigital webbing and fifth finger clinodactyly, hyperextensible hand joints, and shawl scrotum. Short stature was confirmed in 10 cases while the heights of the other six were between the 10th and 25th centile for age. Hair changes were present only in the four originally reported sibs, which might suggest that they are segregating as an independent trait in that branch of the family. None of the 16 cases showed physical disability or mental retardation. Their parents were all phenotypically normal and were either first or second cousins.

This study further characterises the phenotype and confirms the previously suggested autosomal recessive inheritance in this type of faciodigitogenital syndrome. It also highlights the importance of diagnosing such a disorder for proper counselling, particularly in Arab countries with large Bedouin communities such as Kuwait, Saudi Arabia, Qatar, and the United Arab Emirates. It is possible that the gene founder effect originating in this tribe might spread through intermarriage to other Bedouin tribes and probably to the whole population, especially after the recent trend of breaking the traditional practice of consanguineous marriage.

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The two affected males.

1 Teebi AS, Naguib KK, Al-Awadi SA, Al-Saleh QA. New autosomal recessive faciodigitogenital syndrome. *J Med Genet* 1988; 25: 400-6.

2 Farag TI, Teebi AS. High incidence of Bardet-Biedl syndrome among the Bedouin. *Clin Genet* 1989; 36: 463-5

Pericentromeric heterochromatin of chromosome 3

Pericentromeric heteromorphism of human chromosome 3 is usually identified as an intensity variation by the QFQ technique. We show that the *Alu*-Giemsa technique is a much more informative approach.