everyone involved in counselling them has to be sensitive to their beliefs and adherence to Islam. To our knowledge, HD has not been previously recorded in Saudi Arabia, nor in Afghanistan. Although European ancestry was not reported in any family, the disease gene could well have been introduced to them in the last century by foreigners visiting Red Sea or Arabian Gulf ports and, in the case of Afghanistan, travelling along traditional trade routes. Studies have shown that even the most isolated population affected by HD might have had European visitors in previous centuries. At present, the prevalence of HD in this population cannot be estimated, but it is likely to be comparatively low.

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The Prader-Willi-like phenotype in fragile X patients: a designation facilitating clinical (and molecular) differential diagnosis

We thank Gillessen-Kaesbach and Horsthemke for their comment on our paper “Clinical and molecular studies in fragile (X) patients with a Prader-Willi-like phenotype” in the March 1994 issue. The use of “like” syndromic designations generally invokes discussion. We nevertheless chose to describe this rare phenotype of the fragile (X) syndrome in direct comparison with the Prader-Willi syndrome (PWS). The eight fragile (X) patients described in our paper showed a special subphenotype consisting of mental retardation, a full, round face, truncal obesity, hypogonadism, small, broad hands and feet (all prominent PWS symptoms) and regional skin hyperpigmentation. Moreover, they lacked major features of the Martin-Bell phenotype described in the majority of fragile (X) patients: a long face with large everted ears and megaloeutates. Therefore the suggestion by Gillessen-Kaesbach and Horsthemke that these eight patients are just obese fragile (X) patients would be an insufficient clinical description of this special subphenotype. Considering the erroneous clinical diagnosis of PWS in two patients, the (partial) resemblance to classical PWS, and the necessity for a recognisable name for this special subphenotype we suggested the name “Prader-Willi-like”. We hope that this will make clinicians aware of the need for performing DNA analysis for both chromosome 15q abnormalities and the FMR-1 gene mutations in the mentally retarded patients presenting with either the Prader-Willi syndrome or the Prader-Willi-like subphenotype of the fragile (X) syndrome.

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Arthrogryposis multiplex congenita, renal dysfunction, and cholestasis syndrome

We read with interest the article by Horslen et al. “Liver histology in the arthrogryposis multiplex congenita, renal dysfunction, and cholestasis (ARC) syndrome: report of three new cases and review.” J Med Genet 1994;31:624–31. We also support the suggestion that this syndrome is more common than realized and should be considered in the differential diagnosis of cholestasis and renal dysfunction in infants.