
Supplementary Note. Further clinical notes and photographs.

All samples were obtained through the X-linked mental retardation (XLMR) study at the Greenwood Genetic Center. Informed consent was obtained from all families which included specific permission to publish photographs. XLMR research at the Greenwood Genetic Center has been approved by the Institutional Review Board (IRB) at Self Regional Hospital in Greenwood, SC.

Clinical details on the four kindreds with multiple affected individuals are given below. Facial photographs of one individual each from K8610 and K8675 are also shown.

Kindred 8610 is a Caucasian family with two affected males in two generations (Graham et al., 1998; Graham et al., 1999). Only limited details are available on a third male who died at four days with a heart defect. The two living males are undergrown and have high prominent foreheads, small simple ears, wide flat thumbs and great toes, horizontal palmar creases, cryptorchidism, constipation, dysgenesis of the corpus callosum, hypotonia progressing to spasticity, and behavioral disturbances. Macrocephaly and inguinal hernia occurred in III-3; hypertelorism, downslanting palpebral fissures, short hands and feet, and seizures occurred in IV-1. IV-1 had an IQ of 50; III-3 an IQ of
<20. IV-1 had duplication of the great toes and split right hand.

Kindred 8675 is an African American family with two affected males in two generations. A third probably affected male died at three weeks with imperforate anus and heart defect, but no further details are available. The two living males have high prominent forehead, small simple ears, wide flat thumbs, imperforate anus, dysgenesis of the corpus callosum, and mild to moderate mental retardation. One had hypertelorism, hypotonia, and behavioral disturbances. His nephew had relative macrocephaly, frontal hair upsweep, downslanting palpebral fissures, heart defect, inguinal hernia, and seizures.

Kindred 9073 is a Caucasian family with four affected males in three generations. Three males are deceased and significant clinical information is not available on them. The surviving male has relative macrocephaly (OFC 75th centile, height 20th centile), high
prominent forehead with frontal hair upsweep, downslanting palpebral fissures, epicanthus inversus, detached retina, strabismus, small simple ears, open mouth, high narrow palate, microretrognathia, undefined cardiac murmur, wide flat thumbs, hypotonia, dysgenesis of the corpus callosum, dystonia of the neck, severe mental retardation, and aggressive and compulsive behavior.

Kindred 9346 is a Caucasian family and is the original family to be reported with FG syndrome. In the first report, five males, all cousins, were affected (Opitz and Kaveggia, 1974). Later, an additional affected male was born into the family (Riccardi et al. 1977). Details presented here relate only to the first five affected males in the original report, only one of whom survives. The most common findings were hypotonia (5/5), imperforate anus or other anal anomalies (4/5), high prominent forehead (4/5), and relative macrocephaly (3/5). Other findings were recorded less commonly, but include agenesis of the corpus callosum, short stature, frontal hair upsweep, hypertelorism, downslanting palpebrae, small simple ears, short hands and fingers, wide flat thumbs, horizontal palmar creases, heart defect, cryptorchidism, and inguinal hernia. Mental retardation ranging from mild to severe was present in all four who lived beyond the newborn period.