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Partial deletions of chromosome 13 are rare. Based on clinical features, these deletions have been categorized into three groups [Brown et al., 1993, 1995]. Group 1 deletions comprise the chromosome region proximal to 13q32 and are characterized by mild to moderate mental retardation and variable facial dysmorphic features. Patients with deletions of the RB1 locus in 13q14 also have retinoblastoma [Lohmann and Gallie, 2004]. Deletions of 13q32 (Group 2) are associated with the most severe phenotype, often including malformations of the brain, eyes, distal limbs, and the genitourinary and gastrointestinal tract.

Four of the six males listed in Table I were reported to have genital abnormalities, comprising penoscrotal transposition plus hypospadias (our Patient 4), “ambiguous genitalia” (not specified in detail) [Luo et al., 2000], isolated hypospadias [Turleau et al., 1978], and cryptorchidism (our Patient 2).