Concerning the Article by Ogata et al. [1]

Hypergonadotrophic Hypogonadism in a 3-Year-Old Girl with Blepharophimosis, Ptosis, and Epicanthus inversus Syndrome

In the September issue of Hormone Research, Ogata et al. [1] describe a prepubertal patient with blepharophimosis, ptosis, and epicanthus inversus syndrome (type 1 BPES) with hypergonadotropic hypogonadism. Although the syndrome is subdivided into type 1 with infertility of affected females and type 2 with male and female autosomal dominant transmission, it is not known whether the two forms are allelic variants [2]. Both types are heterogeneous and have been linked to a 3q22–23 continuous gene deletion with larger deletions having more dysmorphic features, short stature, and mental retardation [2].

Other endocrine abnormalities are not frequent, although the single mesial maxillary tooth that is sometimes associated with growth hormone deficiency and the holoprosencephaly sequence were described by Karimi-Nejad et al. [3] – in that case computed tomography scans of the brain were normal. I here describe an 11.4-year-old male with type 2 BPES probably inherited from his father who presented with a height SD score of −1.52 as compared with a mid-parental height of +0.4. The TW2 bone age was 9.5 years. The peak growth hormone level to stimulation was 6.5 mIU/l; normal lutestimining hormone releasing hormone test (luteinizing hormone 0.3, 2.1, and 2.7 mIU/ml and follicle-stimulating hormone 2.6, 5.6, and 8.3 mIU/ml at 0, 20, and 60 min, respectively) with normal thyrotrophin-releasing hormone, prolactin, and cortisol response. Karyotype normal and chromosome 3 normal to detailed banding studies. Magnetic resonance imaging showed hypoplastic anterior pituitary with normal stalk.

Apparently isolated growth hormone deficiency was present in this case of BPES associated with a small pituitary gland and presumably only a small, undetected, gene deletion. Many genes are implicated in normal pituitary development and may be associated with midline facial and eye abnormalities [4]. It is possible that some cases of BPES may have pituitary abnormalities accounting for short stature and that chromosome 3 may be worth searching for further genes related to pituitary development.

References