A Case of XYY Syndrome with Short Stature


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APPROXIMATELY 1 in 1000 newborn males displays an XYY chromosome pattern [1]. The 47, XYY male has few phenotypic manifestations. XYY males tend to be tall and are often afflicted with severe nodulocystic acne [1]. Clear-cut endocrine abnormalities have yet to be established. It is not certain why XYY individuals are more apt to be found in mental or penal institutions although it is possible that an abnormality of neural development due to the XYY genotype favors deviant behavior in some. The nature and extent of such an association are yet to be determined. Our patient, diagnosed as having XYY syndrome, was characterized by short stature. We examined the endocrinological status and genetic defects of his Y chromosome.

Case Report

This 8 3/12-year-old boy is the first of 2 children; his younger brother is healthy. He was born at 42 weeks gestation to a 25-year-old mother (153 cm) and a 34-year-old father (162 cm) after an uneventful pregnancy and delivery, with normal Apgar scores. He weighed 3020 grams.

His academic performance in school was poor and he was diagnosed as suffering from attention deficit hyperactivity disorder at the Kyoto City Child Welfare Center. Chromosome analysis revealed that the subject had XYY syndrome (Fig. 1). He was then prescribed a central nervous stimulant (methylphenidate) for 1 year, followed by a major tranquilizer (haloperidol) for the next 3 months. He was referred to our hospital for examination of his characteristic short stature.

His height was 113.6 cm (-2.6 SD) and his bone age was 6.4 years (B.A./C.A.=0.78). He had no pubic hair and his testes were 2.5 ml, which is normal for his age. He displayed neither minor anomalies nor nodulocystic acne.

Routine hematological and biochemical readings

Fig. 1. Chromosome analysis showed that the subject had XYY syndrome.
were all within normal limits (Table 1). His plasma insulin-like growth factor I (IGF-I) had decreased slightly (83.4 µg/L) and the maximal GH responses to insulin, arginine, levodopa and GH releasing factor (GRF) were 13.4, 21.4, 32.4, and 11.1 µg/L, respectively (Fig. 2). For further examination of the GH-IGF axis, we measured IGFBP-3, total IGF-I, free IGF-I and IGFBP-3 protease (Table 1). Because these data were all within the normal range, we concluded that his GH-IGF axis was normal.

Analysis of his Y-specific growth gene revealed no deletion. Other endocrinological examinations revealed no abnormal findings (Table 1). On loading with LHRH and TRH, LH, FSH, TSH indicated normal responses. Brain magnetic resonance imaging revealed abnormal findings in neither the hypothalmus nor pituitary gland.

### Discussion

The XYY male is usually tall [1], but our patient was of short stature (-2.6 SD), and was devoid of any abnormalities in biochemical and endocrinological findings. His GH secretion test, IGF-I, IGFBP-3 and IGFBP-3 protease were all within the normal range [2], indicating that the GH-IGF-I axis was normal. Since his parents are not short, familial short stature was discounted. On the other hand, the presence of a Y-specific growth gene has been postulated [3]. According to this theory, tall stature is induced when the number of Y-specific growth gene increases. It therefore appeared that he may have an abnormal Y chromosome, where the Y-specific growth gene is abnormal, but no deletion on the Y-specific growth gene region was observed.

Individuals with Kleinfelter’s syndrome are usually tall with long extremities, but there have been some conflicting reports describing such patients with short stature [4, 5]. Chromosomal disproportion may play a role in the characterization of his short stature [6]. The short stature encountered in our study might be induced by a disorder in either the post IGF-I receptor mechanisms or
manifestation of the Y-specific growth gene. There is a possibility that he had a constitutional short stature. Otherwise, another unknown factor that regulates growth may be irregular in this case.

References