Early Infantile Overgrowth due to XYY Syndrome

Hiroaki Takahashi, Keiso Tachi, Yuko Fujisawa, Mashio Kitatani, Mamoru Ozaki and Fukumi Kitamoto

Department of Pediatrics (H.T., K.T., Y.F.), Division of Clinical Genetics (M.K., M.O.), Department of Psychiatry (F.K.), Kanazawa Medical University, Ishikawa, Japan

Abstract. A 20-months-old boy was referred for evaluation of his growth and development. Past history revealed that he was the first offspring of a healthy mother at full term and by normal delivery (3,600 g in weight and 52 cm in length). His early milestones were normal, but overgrowth was already noted in early infancy. He weighed 9.5 kg and was 68 cm tall at four months of age. On physical examination at 20 months, he weighed 17 kg (+4.22 SD) and was 88.4 cm tall (+2.05 SD) with a head circumference of 51.4 cm (>2 SD). He was somewhat hyperactive, but did not have temper tantrums or show aggressive behavior. His psychomotor development was delayed with a DQ of 62. EEG showed abnormal basic activity. Chromosome analysis revealed XYY mosaicism. The case demonstrated overgrowth of early infantile onset due to XYY syndrome.

Key words: XYY syndrome, overgrowth, tall stature

Introduction

Despite an incidence of one in 840 newborn males, the XYY syndrome is seldom detected during childhood or even in the adult [1,2]. We have recently observed a case of this syndrome who showed extreme overgrowth of infantile onset with delay in psychosocial development.

Case Report

The case, N.K., a 20-months-old boy, was referred to us for evaluation of his growth and development. Past history revealed that he was the first offspring of a 20-years-old healthy mother (163 cm in height) at full term and by normal delivery. He weighed 3,600 g (+0.98 SD) and was 52 cm in length (+1.10 SD) at birth. His mother denied previous miscarriages. His father (167 cm in height) was 38 years old at the time of his birth. Family history revealed no relative with extreme tall stature or mental retardation, and consanguineous marriage was denied. His early milestones seemed to be normal; however, overgrowth was already noted in early infancy. He weighed 9.5 kg (+2.58 SD) and was 68 cm in length (+2.23 SD) at four months of age (Fig. 1). He had frequent ear and skin infections, but his general health had
been satisfactory. His gross motor milestones seemed to be normal, but his speech was delayed. He did not have temper tantrums or show aggressive behavior.

On physical examination at 20 months of age, he weighed 17 kg (+4.22 SD) and was 88.4 cm in length (+2.05 SD) with a head circumference of 51.4 cm (>2 SD). He showed a robust physique without coarse features and he was slightly hyperactive. He had mild genu varum and multiple skin infection scars with pigmentation, but no café-au-lait spot was noted. He had otitis media with hearing difficulty by auditory brainstem response. His psychomotor development was delayed, with a total DQ of 65 on the Kinder infant development scale and an IQ of 62 by the Tanaka-Binet test (Fig. 2). Chromosome analysis revealed 46,XY (80%)/47,XYY (20%). Bone age was interpreted as 2 3/12 years at the chronological age of 1 8/12 years, by the Greulich and Pyle method. Laboratory data showed normal blood count and chemistry.

**Fig 1.** Growth chart of the XYY patient

<table>
<thead>
<tr>
<th>Areas Evaluated</th>
<th>D AQ</th>
<th>D Q</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Gross Motor Skill</td>
<td>1:10</td>
<td>59</td>
</tr>
<tr>
<td>2. Fine Motor Skill</td>
<td>2:2</td>
<td>70</td>
</tr>
<tr>
<td>3. Language Comprehension</td>
<td>2:2</td>
<td>70</td>
</tr>
<tr>
<td>4. Language Expression</td>
<td>2:0</td>
<td>65</td>
</tr>
<tr>
<td>5. Cognition</td>
<td>2:0</td>
<td>65</td>
</tr>
<tr>
<td>6. Interaction with Children</td>
<td>1:10</td>
<td>59</td>
</tr>
<tr>
<td>7. Interaction with Adults</td>
<td>2:2</td>
<td>70</td>
</tr>
<tr>
<td>8. Self-Control</td>
<td>2:2</td>
<td>70</td>
</tr>
<tr>
<td>9. Eating Behavior</td>
<td>1:10</td>
<td>59</td>
</tr>
</tbody>
</table>

**Fig 2.** Psychometric profile of the XYY patient
Infantile Overgrowth due to XYY Syndrome

Endocrinological studies were within the normal range. Screening test for aminoaciduria and organic aciduria was negative. Electroencephalogram showed abnormal background activity and occiput-dominant high voltage slow waves. NMR-CT scan demonstrated delayed myelinization and a high signal area in the periventricular posterior horn, which might suggest perinatal ischemia.

Discussion

Valentine et al estimated an incidence of one in 846 newborn males for the XYY syndrome [1,2]; however, the XYY individual is seldom detected during infancy or childhood. The features of the XYY syndrome are often subtle and not overtly suggestive of a chromosome abnormality disorder. On the other hand, among institutionalized male juvenile delinquents, the incidence of XYY was 1/35, which is 24 times the frequency in newborn males. Behavioral problems, especially temper tantrums and aggressive or defiant activity, start in early childhood and are sometimes augmented by mental retardation. Witkin et al indicated that the high crime rate for XYY syndrome patients was mainly due to their dull mentality rather than their aggressiveness [3].

Most of the adult XYY cases reported were extremely tall; however, the average final height for XYY syndrome in the Japanese population is not yet available [1-8]. It has been said that the tendency toward tall stature in XYY syndrome is not evident until five to six years of age, although it is occasionally present at birth. However, two of four XYY infants reported by Valentine et al. [1] were at or above the 97th percentile at 9 months of age. The XYY case reported by Miyamoto et al. [9] was born at 39 weeks of gestation, weighed 2,380 g (small for date) and was 45.7 cm in length. At two years and eight months, he weighed 20.2 kg (+5.1 SD) and was 93.4 cm in length (+0.8 SD), and the early infantile overgrowth of XYY syndrome was well documented. The XYY case reported here was long at birth and showed excessive growth starting in early infancy without any significant features of chromosome abnormality. However, chromosome analysis demonstrated XYY mosaicism. He had slightly advanced bone age. Classical cerebral gigantism (Sotos syndrome) may also show excessive prenatal growth and overgrowth in infancy, but they may not end up with tall adult stature, because of advanced bone age [10]. It therefore appeared to be very difficult to predict the ultimate adult height in the present XYY case.

This case demonstrated early infantile onset overgrowth due to the XYY syndrome. Prenatal and postnatal overgrowth might be an indication for chromosome analysis, as this XYY patient shows.

References


