A New Overgrowth Syndrome with Megaepiphysis and Hypertrophy of the Mitral Valve

Seiji Sato, Nobutake Matsuo, Masahiro Banba and Gen Nishimura

Departments of Pediatrics and Diagnostic Radiology, Keio University School of Medicine

Abstract. We report the case of a 2.5-year-old Japanese boy with a new overgrowth syndrome who presented with excessive postnatal growth, mental retardation, unusual facial features, megaepiphyses in the long bones and hypertrophy of the mitral valve. The facial features included a prominent forehead, supraorbital ridges, hypertelorism and a long philtrum. The urine mucopolysaccharides and leucocyte morphology were unremarkable. This pattern did not fit any known overgrowth syndrome.

Key words: overgrowth, megaepiphysis, hypertrophic mitral valve, abnormal facial features

Various overgrowth syndromes have been defined, including Beckwith-Wiedemann syndrome, Sotos syndrome, Weaver syndrome, Marshall-Smith syndrome and Moreno syndrome [1-6]. None, however, is characterized by diffuse megaepiphyses of the long bones. We report here a new overgrowth syndrome manifesting increased linear growth in infancy, megaepiphyses and hypertrophy of the mitral valve.

Case Report

A boy was born after an uncomplicated 40 week pregnancy and delivery to a 38 year old mother. The parents were both healthy and unrelated. A four year old elder brother was normal. There was no family history of malformation, mental retardation or inherited metabolic disease. At birth he weighed 3260 g and measured 51 cm. He was first seen in our clinic for developmental delay at 21 months of age at which time he did not utter a meaningful word and was unable to walk.

Physical examination: At age 21 months, he weighed 12 kg (75th percentile) and measured 90 cm (>97th percentile). His head circumference was 50 cm (97th percentile). Abnormal facial features included a prominent forehead, supraorbital ridges, hypertelorism, a long philtrum and a high arched palate (Fig. 1). A grade 3 continuous murmur was heard at the upper left sternal border and a grade 2 early diastolic murmur was heard at the left mid-sternal border. There was no hepatosplenomegaly, joint contracture or luxation. His external genitalia were Tanner

Correspondence: Seiji Sato, M.D., Department of Pediatrics, Keio University School of Medicine, 35 Shinanomachi Shinjukuku, Tokyo 160 Japan
stage 1 and the testicular volume was 2 ml by Prader's orchidometer.

**Laboratory data:** The morphology of the white blood cells and bone marrow cells was normal. His other laboratory data including blood GH and somatomedin-C were normal. The urinary hydroxyproline and mucopolysaccharides were not increased. The karyotype was 46,XY.

**Cardiac study:** Chest X-ray showed a normal cardiac silhouette with a cardiothoracic ratio of 62%. ECG showed changes consistent with bilateral ventricular hypertrophy. Echocardiograms demonstrated PDA, PI, thickened ventricular walls and septum, and a moderately hypertrophic prolapsed mitral valve (Fig. 2).

**Radiological study:** A skeletal survey showed megaepiphyses in the long bones (Fig. 3) and a bone age of 42 months at a chronolog-
A New Overgrowth Syndrome

Fig 4. Growth curve

Growth pattern (Fig. 4): Reduced growth was noted in the first two months of life but increased growth became apparent between two and four months. His height exceeded the 97th percentile at 21 months and he continued to grow at or above the 97th percentile.

Discussion

We report a hitherto undescribed overgrowth syndrome. The present case shares certain features with other overgrowth syndromes which usually occur in mentally retarded children, but it is distinguishable by the facial features, megaepiphyses and hypertrophy of the mitral valve. Accelerated growth in infancy and the valvular changes initially suggested the diagnosis of mucopolysaccharidosis. However, at 21 months of age the patient has not begun to deteriorate and had not developed other signs of mucopolysaccharidosis. The cause of this overgrowth syndrome is unknown.

References