This male patient was born to nonconsanguineous Japanese parents at 38 weeks of gestation. His parents and sister were clinically normal. At birth, his body length and weight were – 3 SD from the norm. His expressive language development was severely delayed (only a few words spoken at 5 years of age). At 8 years of age, the patient underwent detailed clinical evaluation for severe short stature. He showed proportionate short stature with a height of 99.6 cm (-4.9 SD) and weight of 13.9 kg (-2.4 SD). He exhibited dysmorphic facial appearance, including a wide mouth, short philtrum, thin vermilion of the upper lip, wide nasal bridge, short palpebral fissures, malocclusion with small teeth, triangular facies, and low-set and posteriorly rotated ears. Physical examinations revealed microcephaly, short neck and small hands and feet. Hand roentgenograms showed severely retarded bone age. Ultrasonography detected no abnormality in the kidney. Biochemical and endocrine studies showed no abnormalities except for hypouricemia caused by hyperuricosuria (serum uric acid, 1.7 mg/dl; normal range 3.7–7.5 mg/dl, fractional excretion of uric acid, 18%; normal range 5–15%). Bone age development markedly accelerated from 9 years of age and reached a nearly adult level when the patient was 13 years of age. His nearly final height at 14 years of age was 137 cm (-3.6 SD). He attended a special educational program in a junior high school and had mild behavior and emotional problems.