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Objectives:

Hypercalcemia, hypothyroidism, and early puberty are the most common endocrine disorders defined in Williams-Beuren syndrome (WBS). Here, endocrine evaluation and long-term follow-up of seven patients with WBS are given.

Methods:

Data were obtained from patient's medical records. WBS was diagnosed by demonstration of the deletion on chromosome 7 by using FISH method (7q11.23). OGTT were performed in four patients. Thyroid ultrasonography was performed. L-T4 was started in patients with

Results:

Six patients were male. Age at diagnosis WBS was 1.04 (3.47) a decimal-year. They all had mild hypercalcemia (9.9-11.1 mg / dL). Three of them had overt hypothyroidism while subclinical hypothyroidism were detected in three patients [(0.66 (5.77) decimal-year]. At the diagnosis, serum TSH was10.5 ± 6.3 µIU / ml and f-T4 was 0.9 ± 0.1 ng / dl. L-T4 was started at 5±3.9 µg / kg. Four patients had thyroid hypoplasia and thyroid agenesis had one. Growth hormone deficiency was determined in one patient. Height SDS was -3.26 at the age of $3^{4/12}$ decimal-years when hGH was initiated and increased to -1.45 at the age of 6.08 decimal-years. Age of onset of puberty in three patients was early according to healthy peer. IGT was detected in three pubertal patients and metformin was started.

Follow-up duration was 5.7 ± 2.1 years. Mean GV was 12.9 ± 7.2 cm and 7.6 ± 2 cm at the end of first and second years of the therapy, respectively. At the last visit, L-T4 dose was 2.9 ± 1 µg / kg. All patients had neurodevelopment retardation and were continuing to special education

Conclusions:

Untreated hypothyroidism also causes mental and motor retardation particularly in infancy period in WBS. IGT could be detected in patients with WBS even if adolescence.

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