61 DECREASED SERUM ADIPONECTIN LEVEL IN OBESE CHILDREN

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Aim: To determine whether serum adiponectin, which is one of the physiologically active gene products secreted from adipose tissue, is decreased in obese children.
Methods: Subjects were 36 consecutive outpatient Japanese obese children, 20 boys and 16 girls, ranging in age from 5 to 14 years, and 24 age-matched nonobese children, 14 boys and 10 girls, as the control group for measuring adiponectin. Blood was drawn after an overnight fast and, at the same time, the obese children were subjected to anthropometric measurements including height, body weight, waist girth, hip girth, and triceps and subscapular skinfold thicknesses. Serum adiponectin was assayed by an enzyme-linked immunosorbent assay kit (Chugai Diagnostics Science Co.).
Results: Serum adiponectin level was lower (6.5 ± 0.7 vs. 10.4 ± 1.0 mg/L, means ± s.e.m., p=0.003) in the obese children than in the controls. In 15 obese children, whose percent overweight declined during therapy, the adiponectin level increased (from 5.5 to 7.0 mg/L, p=0.015). The adiponectin level was correlated inversely with visceral adipose tissue area in obese children. The relationships between adiponectin and other blood biochemistry data were not significant, after being adjusted for either waist girth or body weight.
Discussion and Conclusion: Serum adiponectin level is decreased in obese children depending on the increase in visceral adipose tissue and is restored toward normal level by reducing the degree of obesity.

62 A CASE OF INFANTILE MALIGNANT OSTEOPETROSIS TREATED WITH ALLOGENEIC BONE MARROW TRANSPLANTATION

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Infantile malignant osteopetrosis is a rare disease representing pancytopenia, hepatosplenomegaly due to defective osteoclast function, and carrying extremely poor prognosis without early treatment by bone marrow transplantation. Herein, we describe a case of infantile malignant osteopetrosis treated with allogeneic bone marrow transplantation.
Case Report: A female infant, born at 38 weeks of gestation as the fourth child of the consanguineous parents, was admitted to our hospital at 26 days of age with feeding difficulty. Her first brother was dead by infantile malignant osteopetrosis despite of the treatment with high-dose 1 α OHD3 therapy and Interferon γ. Her second and third sisters are healthy. She showed marked hepatosplenomegaly. Bone radiography indicated monotonously increased bone density. From these findings, she was diagnosed as infantile malignant osteopetrosis. As in her first brother, she showed increased muscle tonus and macular degeneration.
At 34 days of age, she was medicated with predonisolone for thrombocytopenia. At 46 days of age,1 α OHD3 was also medicated. At 53 days of age, she received bone marrow transplantation from her HLA-identical healthy sister. After 1 month, skeletal X-ray revealed the formation of bone marrow cavity. Her liver size was returned to normal.
Bone marrow transplantation rescued the patient from deteriorated hematopoiesis, hepatosplenomegaly, and monotonously increased bone density, but not from increased muscle tonus and macular degeneration.

63 Sleep apnea in children with achondroplasia and other craniofacial anomalies

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【Introduction】
Sleep apneas are often serous problems in children with craniofacial anomalies.
Sleep apnea is usually obstructive due to anomalous stenosis in the upper airway, but occasionally of central origin as seen in achondroplasia.

【Materials & Methods】
Polysomnographic study (PSG) was performed in 18 children with craniofacial anomalies aged 15 years and under, and the results were compared with those in 37 children without craniofacial anomalies who were suspected to have sleep apnea.
The cases with craniofacial anomalies consisted of 6 Achondroplasia, 4 Crouzon, 3 Apert, 2 Treacher Collins ,2 Goldenhar ,and others. PSG was recorded at least twice using Allis 3 (Chest) and the management was decided with reference to our sleep apnea scale on Apnea frequency, SPO_2, $EtCO_2$, and behavior pattern.

【Result & Discussion】
Obstructive apnea was more frequent and serious than in children without anomalies and some had central or mixed apneas.
Following treatments were done, tonsill & adenoidectomy (8), craniofacial plastic surgery (3), Home Oxygen or CPAP therapy (5),and so on. Compared with children without anomalies , the improvements of sleep apnea were less dramatic and multidisciplinary approaches were needed.

【Conclusion】
PSG was thought to be essential for the management of sleep apnea associated with cranio-facial anomalies.

64 GNAS1 mutation in a Japanese boy with progressive osseous heteroplasia.

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Background: Progressive osseous heteroplasia (POH) is a rare disease that is characterized by ectopic progressive ossification of skin, muscle, and connective tissue. We report an 11 year-old Japanese boy with POH, a first case among Japanese ethnic background. At the age of 6 years, physical examination of the patient found ossified subcutaneous nodules and plaques on right neck, shoulder, and waist. The right shoulder was predominantly affected. No associated anomalies were detected. The serum calcium, phosphorus, 1,25 vitamin D, and parathyroid hormone level were within normal limited. His parents and relatives had no signs of heterotopic calcification or signs of Albright hereditary osteodystrophy (AHO). **Method:** Patient's and parents' DNAs were extracted from their lymphocytes with donor's agreement and permission for scientific usage. GNAS1 gene was PCR-amplified using exon specific primers and sequenced in standard method. **Results:** Heterozygous 4-bp (GACT) deletion of GNAS1 gene, creating inappropriate stop codon at downstream, was identified in exon 7. However, no mutation was detected in parents' GNAS1 gene. **Discussion/Conclusion:** Identical mutation identified in this patient has already been reported from patients suffering from AHO or POH. Although recent report has released the hypothesis that the POH is inherited through paternal transmission of GNAS1 mutation, this patient indicated POH phenotype with de novo mutation of GNAS1. Analysis of the origin of the mutated allele in this patient might be a clue for unveiling the mechanism by which identical mutation give a rise to different phenotype between AHO and POH.