(P-08)

A Case of Congenital Generalized Lipodystrophy Type 2 with Novel *BSCL2* Gene Mutation

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Congenital generalized lipodystrophy (CGL) is a rare autosomal recessive disorder characterized by generalized absence of adipose tissue, extreme insulin resistance, hypertriglyceridemia, hepatomegaly, hepatic steatosis, and early onset of diabetes. Herein, we described a case with CGL2 due to novel homozygous *BSCL2* gene mutation.

Three years-seven months old girl presented with a general lack of subcutaneous fat, prominent muscular hypertrophy, hollow cheeks, triangular face, acanthosis nigricans in fold areas, especially in the neck-bilateral axilla, hypertrichosis in arms-legs, abdominal swelling due to hepatomegaly, which are characteristic physical findings of CGL. Her parents were first-degree cousins. In laboratory: Glucose 75 mg/dL (70-105), C-peptide 6.8 ng/mL (0.9-4.3), insulin 47.4 µIU/mL (1.9-23), HbA1c 5.2% (4.8-6.0), total cholesterol 132 mg/dL (< 200), and triglyceride 134 mg/dL (< 200). Hyper triglyceridemia was firstly detected at 5 years of age with metformin therapy. Despite taking metformin treatment, the patient's insulin levels increased steadily, and serum AST levels were also elevated. At the age of nine, grade 2 hepatic steatosis with hepatomegaly was detected in ultrasonography.

During follow-up, her HbA1c level has increased to 6.5% at the age of eleven years and three months. The fasting and 2-hour post-OGTT glucose-insulin levels of the patient were 152 mg/dL-158.3 µIU/mL and 209 mg/dL-95.8 µIU/mL, respectively. Insulin detemir was started in addition to metformin treatment because of diagnosis diabetes.

A clinical diagnosis of CGL was corrected by the identification of a novel homozygous mutation (IVS2 + 2 T > C) in the *BSCL2* gene. Analyzes with GenSplicer and Human Splicing Finder modeling programs show that this mutation can cause the disease.

(P-09)

Gene Conversion and Congenital Adrenal Hyperplasia: Two Case Reports

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Congenital adrenal hyperplasia (CAH) is one of the inborn metabolic disorders inherited in an autosomal recessive manner. 95% of CAH cases are due to 21-hydroxylase deficiency. 21-hydroxylase enzyme have an active gene and a pseudogene. The rearrangements between these two genes play an important role in the pathogenesis of CAH. Herein, we present the cases of two siblings with different phenotypes and different chromosomal sex who both have a large gene conversion and a point mutation.

21-hydroxylase gene strip assay and MLPA analysis were performed in the two sibling cases.

The case with male phenotype has been diagnosed with CAH due to salt-wasting crises, macrogenitalia, and hyperpigmentation when he was 1 month old. Karyotype analysis results were 46,XY and SRY(+). The female case with 46,XX has been diagnosed with CAH due to salt-wasting crises and ambiguous genitalia in the newborn period. Results of CAH strip assays were c.89C > T(P30L) (N/M), c.329-336del(Del 8bp E3) (N/M), c.290-13A/C > G (I2 Splice) (M/M) in both cases. In MLPA analysis, heterozygous increase in CYP21A1P-1 (-113 SNP) and CYP21A1-P-3 (del8nt) mutation regions, heterozygous loss in *CYP21A2*-1wt (-113 SNP) and *CYP21A2*-3 wt (del8nt) regions, and homozygous mutations in *CYP21A2*-3 wt (I2 G-C), *CYP21A2*-3 wt (I2G-A) regions were detected. It was thought that the cases have received an allele with heterozygous mutation in c.290-13A/C > G (I2 Splice) region from one parent and a gene converted allele from the other. Mutation analysis was planned for parents.

The cases were presented here in order to emphasize the importance of MLPA analysis when diagnosing CAH.

(P-11)

Heterozygous p.D61G Mutation in a Patient with Noonan Syndrome

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Noonan syndrome is an autosomal dominant disease resulting from mutations in the ras-associated mitogen activating protein kinase pathway involved in signal transduction associated with cell proliferation, differentiation, life, and metabolism.

A girl from non-consanguineous family was referred to pediatric endocrine department because of short stature. The 15-year-old girl was born with weight 2300 g by caesarean section and was followed due to pulmonary valve stenosis and mitral insufficiency in the pediatric cardiology department; she underwent cardiac surgery during the infant period. On physical examination height was 131.6 cm (< 3 p), height SDS -4.73, weight 28.7 kg (< 3 p), weight SDS -5.31, target height 150.65, and target height SDS was 1.95. Physical examination also revealed dysmorphic facial appearance with webbed neck, hypertelorism, epicanthus,