A Case of SHOX Gene Deletion Diagnosed By Microarray

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SHOX (Short Stature Homeobox) which is located at Xp22.33 is evolutionary well-conserved developmental gene expressed in osteogenic cells. SHOX is one of the suspected components of the short stature in Turner syndrome cases. Also functional homolog of SHOX gene is located at Y chromosome. Haploinsufficiency of genes on the X chromosome results in Turner syndrome. Here, we present a 26-month-old female referred to genetic counseling because of short stature and developmental delay. Her height was 71 cm (<3 percentile), weight 9.5 kg (<3 percentile). She had frontal bossing, hypertelorism, and bilateral mesomorphic short upper extremities. Her motor and mental developments were normal. Bone X-ray survey revealed a thickness of long bones and delayed bone age. Karyotype showed an extra genomic material at the p arm of the X chromosome. We performed chromosomal microarray. Approximately 18 Mb gain at the short arm of chromosome 6 and 680 Kb deletion at the p arm of X chromosome were detected. Three genes including SHOX were deleted from the involved region of X chromosome. A gain of 63 genes located at chromosome 6p was observed, which resulted in partial trisomy of 6p. Effects of partial trisomy 6p in our case is not clear, but the deleted SHOX is suspected to be the reason for short stature and delayed bone age.

HOXC4 Gene is Possibly Responsible for Lin-Gettig Syndrome

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HOXC4 (Homeobox C4) is a developmental gene which is expressed in osteogenic cells and is possibly responsible for Lin-Gettig syndrome. This syndrome is described by Lin and Gettig in 1990 as a very rare autosomal recessive disease. The syndrome is characterized by craniosynostosis, severe mental retardation, absence of corpus callosum, dysmorphic facial features, camptodactyly, and hypogonadism. The molecular etiology of the syndrome has not yet been identified. In this report, we present a patient diagnosed as having Lin-Gettig syndrome via clinical findings. Molecular genetic studies have revealed that HOXC4 may be the responsible gene for this syndrome.

POU1F1 and PROP1 Gene Mutations in 4 Cases of Combined Pituitary Hormone Deficiency

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Combined pituitary hormone deficiency (CPHD) is characterized by the impaired production of GH together with one or more of other pituitary hormones. The most commonly recognized genetic defects associated with CPHD include mutations within PROP1, POU1F1, HESX1, LHX3, LHX4, OTX2, GLI2, and SOX3.