



POSTER PRESENTATION

Open Access

Clinical, biochemical, and genetic analysis of two Korean patients with Trichorhinophalangeal syndrome type I and growth hormone deficiency

Young Bae Sohn^{1*}, Chang-Seok Ki², Sung Won Park³, Sung-Yoon Cho³, Ah-Ra Ko⁴, Min-Jung Kwon², Ji-Youn Kim⁴, Hyung-Doo Park², Ok-Hwa Kim⁵, Dong-Kyu Jin³

From 7th APPEs Biennial Scientific Meeting
Nusa Dua, Bali. 14-17 November 2012

Tricho-rhino-phalangeal syndrome type I (TRPSI) is a rare autosomal dominant hereditary disorder characterized by sparse hair, bulbous nose, long philtrum, thin upper lip, and skeletal abnormalities including cone-shaped epiphyses, shortening of the phalanges, and short stature. TRPSI is caused by mutations in the *TRPS1* gene. Herein, we report two Korean cases of TRPSI. Although both patients (a 17-year-old-female and a 14-year-old male) had typical clinical findings, Patient 1 had an additional growth hormone (GH) deficiency. Treatment with recombinant human growth hormone (rhGH) 0.7 IU/kg/week led to an increase in growth velocity. Over 10 years of GH therapy, the mean growth velocity was 5.7 ± 0.9 cm/year. While patient 2 showed a low response after the GH stimulation test, the patient had a poor response with rhGH therapy and GH therapy was discontinued after 6 months.

For the genetic analysis of the *TRPS1* gene, two mutations were found. Patient 1 had a heterozygous mutation c.2520dupT (p.Arg841LysfsX3) which had not been previously reported. Patient 2 had a known nonsense mutation c.1630C>T (p.Arg544X). In summary, we were the first to report Korean patients with mutation of *TRPS1*.

Authors' details

¹Department of Medical Genetics, Ajou University Hospital, Suwon, Korea. ²Department of Laboratory Medicine and Genetics, Sungkyunkwan University School of Medicine, Seoul, Korea. ³Department of Pediatrics Samsung Medical Center, Sungkyunkwan University School of Medicine, Seoul, Korea. ⁴Center for Clinical Research, Samsung Biomedical Research Institute, Seoul, Korea. ⁵Department of Radiology, Ajou University Hospital, Suwon, Korea.

Published: 3 October 2013

¹Department of Medical Genetics, Ajou University Hospital, Suwon, Korea
Full list of author information is available at the end of the article

doi:10.1186/1687-9856-2013-S1-P59

Cite this article as: Sohn et al.: Clinical, biochemical, and genetic analysis of two Korean patients with Trichorhinophalangeal syndrome type I and growth hormone deficiency. *International Journal of Pediatric Endocrinology* 2013 **2013**(Suppl 1):P59.

Submit your next manuscript to BioMed Central
and take full advantage of:

- Convenient online submission
- Thorough peer review
- No space constraints or color figure charges
- Immediate publication on acceptance
- Inclusion in PubMed, CAS, Scopus and Google Scholar
- Research which is freely available for redistribution

Submit your manuscript at
www.biomedcentral.com/submit

