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

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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*.

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