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Schimke immuno-osseous dysplasia 1례

부산대학교 어린이병원 소아청소년과[1], 서울대학교 어린이병원 소아청소년과[2]

김성헌[1], 정해일[2], 김수영[1]

A First Case of Schimke Immuno-osseous Dysplasia in Korea

Seong Heon Kim[1], Hae Il Cheong[2], Su Young Kim[1]

Department of Pediatrics[1], Pusan National University Children's Hospital
Department of Pediatrics[2], Seoul National University Children's Hospital

Introduction: Schimke immuno-osseous dysplasia (SIOD, OMIM #242900) is a rare autosomal recessive disorder consisting of disproportionate short stature from spondyloepiphyseal dysplasia, characteristic face, lymphopenia with defective cellular immunity, progressive renal failure form nephrotic syndrome with FSGS and skin pigmentation

Case: This patient was born at 39 weeks of gestation and delivered via a cesarian section due to oligohydroamnios with a weight of 1.73 kg and a length of 43 cm which were below 3rd percentile. He had experienced many recurrent infections, mostly gastroenteritis and suffered from severe growth retardation since gestation. The patient was referred to our hospital at the age of 4 years and 8 months for persistent proteinuria without significant edema. All body measurements including weight, height and head circumference were below 3rd percentile and he had disproportionate short stature. He had a broad, low nasal bridge and bulbous nasal tip. Physical examination revealed multiple hyperpigmentated macules on his back and very small testis for his age. Skeletal radiographs demonstrated the sign of spondyloepiphyseal dysplasia (SED) characterized by pear-shaped vertebral bodies. Soon after, he developed steroid resistant nephrotic syndrome with normal renal function. Renal biopsy revealed focal segmental glomerulosclerosis (FSGS). Cyclosporine was added to corticosteroid for FSGS treatment, but could not be continued because of leucopenia. On the basis of clinical and laboratory findings, we started to look for the cause of this syndromic FSGS and finally confirmed a first case of SIOD in Korea by genetic analysis, which showed that patient was compound heterozygous for two mutations : a novel missense mutations in exon 9 (c.1484A>C) inherited by the mother and a missense mutation (c.1851+1G>T) in the alternative splicing in intron 12 inherited by the father. After diagnosis of SIOD confirmed, all immunosuppressive agents were withdrawn and he began to be treated with enalapril and simvastatin. After 1.5 year-follow up, nephrotic range proteinuria persists and his renal function is getting worse (estimated GFR 50-60 ml/min/1.73m^2) despite our treatment.

Conclusion: We report the first case of SIOD patient in Korea. Clinical suspicion is important in the case of steroid resistant nephrotic syndrome and growth failure.

Key Words: 쉼케 면역 골이형성증, 첫 증례, 국내
Schimke immuno-osseous dysplasia, First case, Korea