عنوان مقاله:

A Case Report of Schimke Immuno-Osseous Dysplasia: A Rare Autosomal Recessive Disorder

محل انتشار:

مجله بین المللی کودکان, دوره 6, شماره 2 (سال: 1397)

تعداد صفحات اصل مقاله: 5

نویسندگان:

.Banafsheh Arad - *Children Growth Research Center, Qazvin University of medical sciences, Qazvin, Iran*

.Zahra Pirzadeh - *Children Growth Research Center, Qazvin University of medical sciences, Qazvin, Iran*

خلاصه مقاله:

Schimke immune-osseous dysplasia (SIOD) is a rare autosomal recessive disorder presented with specific facial features, skeletal dysplasia, steroid resistance nephrotic syndrome (SRNS) and cellular immune insufficiency. This is a SIOD case reported from Iran. He was 5 years old boy when evaluated for proteinuria and short stature. In appearance, we detected hyperpigmented macules, kyphoscoliosis, and warty lesions. He developed progressive renal failure and steroid resistant nephrotic syndrome, so kidney biopsy was performed and revealed focal and segmental glomerulosclerosis. He didn't respond to prednisolone and Calcineurin inhibitors. He had recurrent lymphopenia with low CD4/CD8 ratio. However lymphopenia respond to granulocyte colony-stimulating factor (G-CSF), he died with pneumonia and sepsis. Nephrotic syndrome due to focal segmental glomerulosclerosis may be accompanied by syndromes. In Qazvin province, we see autosomal recessive disorders more, because of consanguineous marriages. To the best of our knowledge, this is the fourth case of SIOD to be reported from Iran.

کلمات کلیدی:

case report, Child, Lymphopenia, Schimke-Immuno-osseous Dysplasia