Coexistence of Hypomelanosis of Ito and Focal Segmental Glomerulosclerosis: Case Report

Hypomelanosis of Ito (HI) is a neurocutaneous disorder characterized by hypopigmented whorls, streaks and patches with irregular border along the lines of Blaschko. Although central nervous, musculoskeletal and ocular system defects are most frequent complications of HI, renal involvement has been reported less frequently. Five case reports of HI associated with renal diseases including glomerular basement membrane abnormalities, glomerulonephritis, polycystic kidney disease, glomerulocystic kidney disease or focal segmental glomerulosclerosis (FSGS) were defined in the literature. We reported a 4-year-old girl with cutaneous findings of HI who developed proteinuria in the follow-up period consistent with FSGS. Our case is presented to emphasize the necessity of following HI patients more carefully in terms of other organ defects which may develop rarely.

Key Words: Pigmentation disorders; glomerulosclerosis, focal segmental


Anahtar Kelimeler: Pigmentasyon bozuklukları; glomeruloskleroz, fokal segmental


Hyomelanosis of Ito (HI), also termed incontinentia pigmenti ac-
with cutaneous findings of HI who developed proteinuria consistent with focal segmental glomerulosclerosis (FSGS).

**CASE REPORT**

A 4-year-old girl with irregular white spots on her body surface was presented to our department. She was born by caesarian section at 38 weeks gestation and her birth weight had 2900 g. The pregnancy and perinatal periods have lasted uncomplicatedly. Two months after birth, a hypopigmented rash was noted on her right leg, subsequently new lesions with irregular shape and size were seen on her extremities and trunk in the following two years. The abnormal skin lesions persisted throughout her childhood.

Cutaneous examination revealed hypopigmented maculae with linear streaks and whorls involving the trunk and extremities (Figure 1). General physical examination revealed only strabismus. The diagnostic tests for identifying another organ involvement included; routine hematological, biochemical and urinalysis tests, cranial computed tomography, radiographs of the chest and extremities, abdominal ultrasound, electrocardiography and echocardiography were performed. The only abnormality detected was cerebellar hypoplasia screened by cranial computed tomography. Neurologic examination was normal except for mild dysmetria at tandem walking; this finding was not consistent with HI. Denver Developmental Screening Test also indicated normal findings in all developmental parameters. A genetic examination, yielding normal results (46,XX), was performed to exclude a chromosomal deviation.

During follow up, persistent nephrotic proteinuria (47.4 mg/m²/h) with a normal glomerular filtration rate were documented. Abdominal ultrasound showed grade 1 hypeeregenic kidneys. A percutaneous kidney biopsy was performed and light microscopy showed segmental sclerosis in 4 of 15 glomeruli with moderate interstitial fibrosis and mild tubular atrophy consistent with FSGS (Figure 2). Following histopathologic diagnosis captopril treatment was initiated.

**DISCUSSION**

Hypomelanosis of Ito is a neurocutaneous disorder including pigmentary anomalies and defects of other organs. The skin lesions of HI appear at or shortly after birth, in about 70% of patients within
the first year of life. The lesions may be unilateral or bilateral and appear anywhere on the body with the exception of the palms, soles, scalp, and mucous membranes.¹

Excutaneous abnormalities have been demonstrated in about 75% of HI patients. The most frequent complications are related to central nervous, musculoskeletal and ocular system involvement. Central nervous system disorders consist of mental retardation, epilepsy, ataxia, autistic-like behavior and electroencephalographic changes. Cranial computed tomography and magnetic resonance images can demonstrate focal or generalized cerebral atrophy, white matter changes and cerebellar atrophy or hypoplasia. Musculoskeletal defects include scoliosis, thoracic deformity, hypotonia, limb and finger anomalies. Ophthalmological abnormalities consist of strabismus, nystagmus, myopia, cataract, retinal degeneration and optic atrophy. The spectrum of abnormalities in other organ systems are hair, teeth, nail, sweat gland, craniofacial, congenital cardiac and gastrointestinal defects.¹²

Renal involvement has been reported only exceptionally. Only few cases of HI with renal disease described in the literature.³⁷ Chevalier et al. described a 4-year-old male of HI with glomerulonephritis and glomerular basement membrane abnormalities, as the first report.³ Subsequently, a male patient with HI, tuberous sclerosis, polycystic kidney disease and α-thalassemia trait was reported by Eussen et al.⁴ The authors speculated that these findings could be at least partly explained by a contiguous gene syndrome (e.g. affecting loci on chromosome 8 and 16) and either chromosome might contain a “HI” gene.⁴ In addition, a 13-year-old girl of HI with structural anomalies of the kidneys, including macroscopic and microscopic cysts, foci of tubular atrophy and interstitial fibrosis was reported by Coward et al.⁵ Recently, Vergine et al. described a male infant with a glomerulocystic kidney disease with HI.⁶ The association of HI and FSGS has been previously reported by Gatter et al. in an 8-year-old girl who developed proteinuria. She had also deafness and skeletal defects such as short stature, scoliosis, finger anomalies and facial asymmetry.⁷ In our patient, who had cerebellar atrophy and strabismus, proteinuria developed in the follow-up period and treatment was initiated immediately following histopathological diagnosis. Determination of proteinuria is regarded as coincidentally because she was asymptomatic. However, early diagnosis and treatment might prevent renal failure due to FSGS in our patient.

In conclusion, we believe that HI might be a hallmark of a wide range of concomitant alteration of the different organ systems. Therefore, the patients of HI should be followed more carefully in terms of other organ defects which may develop less frequently.

REFERENCES