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Overview of congenital cataracts with genetic defect on the occasion of a rare case

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Cataract which means clouding in eye lens may result in a wide variety of visual outcomes from blurry vision to blindness. Moreover, if cataract has congenital roots, it may turn out to be a huge power affecting the course of whole life in patients as visual development will be deeply affected. Therefore, it is important whether timely and appropriate treatment is performed. Congenital cataract occur about 3 in 10000 live births. Besides, cataracts coupled with genetic abnormalities are much rarer. In this presentation, we aimed to draw attention to syndromic cataracts by reporting a rare instance of congenital cataract with Hallermann-Streiff syndrome (HSS). One-month-old full-term female infant has been referred to our clinic since the cataract etiology cannot be determined. Examination showed bilateral congenital cataract, microphthalmia and microcornea. Patient's medical history was normal except parents were relatives. We didn't find any abnormalities that can shed light on etiology of cataract in the systemic investigation including TORCH panel. When we re-evaluate the patient in detail, we noticed bird-like face, dental abnormalities, hypotrichosis, skin atrophy and short stature. In the light of these findings, the patient was diagnosed with HSS. HSS diagnosis was confirmed by chromosome analysis which showed gene defect in sixth chromosome (6q21-q23.2). Bilateral limbal lensectomy was performed for the cataract treatment. In conclusion, HSS is a rare condition that possesses a typical facial appearance. True diagnosis of congenital cataracts with unclear etiology must be made by thoroughly evaluating the cases and recording any atypical phenotypic finding.

Biography

Hanife Tuba Akcam has completed her MD at Kırıkkale University and Residency at Gazi University School of Medicine. She is an Assistant Professor at Duzce University Medical School. She has published various papers in reputed journals. Also, she has a lot of project about pediatric eye disorders.

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