Hereditary Hyperferritinemia Cataract Syndrome: Case Report

Kalıtsal Hiperferritinemi Katarakt Sendromu: Olgu Sunumu

Zeliha Guzelkuçuk [1], Hasan Fatih Cakmaklı[2], Mehmet Onen [3], Antonello Pietrangelo [4], Nese Yarali[5]

[1] University of Health Sciences, Ankara City Hospital, Department of Pediatric Hematology and Oncology, Ankara, Turkey
[2] Ankara University, Faculty of Medicine, Department of Pediatric Hematology and Oncology, Ankara, Turkey
[3] Numune Training and Research Hospital, Department of Ophthalmology, Ankara, Turkey
[4]University Hospital of Modena, Unit of Internal Medicine and Metabolic Diseases Center for Hemochromatosis, Modena, İtalya
[5]Yıldırım Beyazıt University, Ankara City Hospital, Department of Pediatric Hematology and Oncology, Ankara, Turkey

ABSTRACT

Hereditary Hyperferritinemia-Cataract Syndrome (HHCS) is a rare disease characterized by cataract and hyperferritinemia. Herein, we present a pediatric patient diagnosed with HHCS.

Key Words: Hyperferritinemia , cataract, L-Ferritin

Received: 06.02.2020 **Accepted:** 05.11.2020

ÖZET

Kalıtsal Hiperferritinemi-Katarakt Sendromu (HHCS) katarakt ve hiperferritinemi ile karakterize nadir bir hastalıktır. Burada, HHCS tanisi alan çocuk hasta sunulmaktadir.

Anahtar Sözcükler: Hiperferritinemi, katarakt, L-Ferritin

Geliş Tarihi: 06.02.2020 **Kabul Tarihi:** 11.05.2020

ORCID IDs: Z.G. 0000-0003-1462-6867, H.F.C. 0000-0003-0152-1564, M.O.0000-0001-8797-9634,A.P. 0000-0002-7411-935X, N.Y.0000-0001-5488-2385

Address for Correspondence / Yazışma Adresi: Zeliha Guzelküçük, MD Ahi Evran University Research and Training Hospital, Kirsehir, Turkey E-mail: drguzelkucuk@hotmail.com

doi:http://dx.doi.org/10.12996/gmj.2020.108

INTRODUCTION

Hereditary Hyperferritinemia-Cataract Syndrome (HHCS) is a rare disease with autosomal dominant inheritance and characterized by cataract and hyperferritinemia (1). The diagnosis of HHCS is easy in case of hyperferritinemia, presence of cataract, family history, normal serum iron and transferrin saturation. Herein, we presented a rare case of HHCS in a child.

CASE REPORT

An 11-year-old female patient was admitted to pediatric hematology department due to high levels of serum ferritin. The patient had no complaints other than a decrease in visual functions that had recently become apparent. She had no history of rheumatic or infectious disease. Red blood cell transfusion was not received. Her mother, two aunts and grandmother had cataract operation at early ages.There was no family consanguinity. Physical examination was normal. Complete blood count, blood electrolytes, renal and liver function tests and blood lipid levels were normal. Serum ferritin level was 1111.1 ng / ml (normal range: 11-306). C-reactive protein values were within normal limits. Serum iron level was 43 µg / dL (normal range: 37-145), iron binding capacity was 325 µg / dL (normal range: 110-370) and transferrin saturation was 13.2%. T2* cardiac and R2 * liver magnetic resonance imaging (MRI) for the detection of iron overload findings revealed no iron deposition in the heart and liver. Visual acuity was measured as 0.6 in the right and left eyes on ophthalmologic examination and cataract and visual loss were detected. It was thought to be compatible with HHCS because the cataract was typically composed of opacities located in the lens cortex (Figure 1). Genetic testing was performed with the preliminary diagnosis of HHCS due to history of familial early-onset cataract and hyperferritinemia. The diagnosis was confirmed. (Ferritin L UTR: FTL 5'UTR, 40A>G). Cataract surgery was not performed at this stage and it may be needed during follow-up. The patient was followed up by the ophthalmology department for ocular findings.

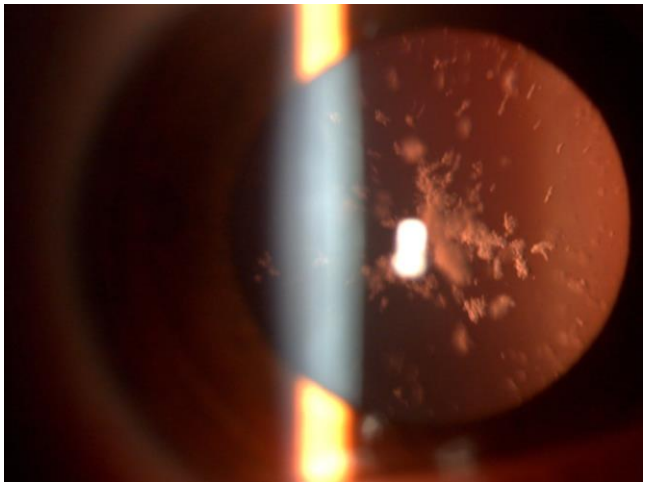

Figure 1. Biomicroscopic view of cataract associated with hyperferritinemia with multiple small lens opacities located in the lens cortex

DISCUSSION

Ferritin; is the main storage protein of non-ferrous iron in the body. It consists of two chains, heavy and light chains (2,3). As a result of the heterozygous mutation of the iron-responsive element (IRE) on the light chain gene, the regulation of ferritin synthesis is influenced, and the light chain synthesis, defined as L-ferritin, becomes continuous (3,4). This may cause increased serum ferritin levels. Increased L-ferritin settles in the lens and causes early onset bilateral cataract (5,6). It can be distinguished from other causes of cataract with its pathognomonic appearance, which is mostly located in the lens cortex and consists of several small opacities. Among these small opacities, the lens is viewed transparent. Electron microscopic studies on lens opacities have shown that these opacities are ferritin deposits that cause scattering of light (6). Typical ophthalmic findings were also detected in our patient. Other family members of the patient were also examined and ferritin levels were measured. Her mother, grandmother, and two aunts were affected by early-onset cataracts and hyperferritinemia were also found in them.

Ferritin does not accumulate in organs other than the lens in HHCS. Liver, heart and other organs are protected from damage of ferritin accumulation (5). Therefore, invasive diagnostic procedures are not necessary. Cataract surgery should be performed only at the appropriate age for treatment. Bilateral pathognomonic ophthalmic involvement is typical and adult patients seem to tend to have slow progression (7).

CONCLUSION

HHCS is a rare disease with early onset cataract and high ferritin levels, and normal TS. Detailed family history could protect patients from unnecessary invasive procedures.

Conflict of interest

No conflict of interest was declared by the authors.

REFERENCES

1.Yin D, Kulhalli V, Walker AP. Raised serum ferritin concentration in Hereditaryhyperferritinemia cataract syndrome is not a marker for iron overload. Hepatology, 2014 ;59:1204-6.

2.Taner Özgürtaş, İbrahim Aydın, A. AvniAtay at al. Yüksek Serum Ferritin Düzeylerinin Etiyolojik Spektrumu, Turkish Journal of Biochemistry–Turk J Biochem, 2008; 33; 223–5.

3.Tsantoula F, Kioumi A, Germenis AE, at al. Hereditaryhyperferritinemia cataract syndrome as a cause of childhood hyperferritinemia. J PediatrHematolOncol, 2014;36:e304-6.

4. Perruccio K, Arcioni F, Cerri C, at al. The hereditary hyperferritinemia-cataract syndrome in 2 italian families. Case Rep Pediatr, 2013;2013

5. Bowes O, Baxter K, Elsey T, at al. Hereditary hyperferritinaemia cataract syndrome. Lancet, 2014. 26;383:1520

6. Yazar S, Franchina M, Craig JE, at al. Ferritin light chain gene mutation in a large Australian family with hereditary hyperferritinemia-cataract syndrome. Ophthalmic Genet, 2017;38:171-4.

7. Cosentino I, Zeri F, Swann PG, Majore S, at al, Grammatico P,Petitti V. Hyperferritinemia-cataract syndrome: Long-term ophthalmic observationsin an Italian family. Ophthalmic Genet, 2016 ;37:318-22.