Case Report

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A Case Report of Down Syndrome and β-Thalassemia Major Coincidence

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Received: 06 July 2023 **Revised:** 10 August 2023 **Accepted:** 22 August 2023

ARTICLE INFO

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Keywords:
Down syndrome;
Beta-Thalassemia; Transfusion Therapy;
Karyotype

ABSTRACT

Background: Down syndrome and β-thalassemia are commonly prevalent genetic diseases worldwide. Predominantly, an extra copy of chromosome 21 or trisomy 21 predominantly causes Down syndrome (the most common genetic etiology of moderate intellectual disability). Down syndrome is associated with congenital anomalies and characteristic features. β-thalassemia major or transfusion- dependent thalassemia refers to a severe expression of the disorder that requires early transfusion therapy.

Case Report: Here, we reported a male Down syndrome patient with a 47, xy, +21 karyotype who was diagnosed with β-thalassemia major at 6 months and treated with repeated transfusions every 20 days due to anemia.

Conclusion: The association between Down syndrome and major β-thalassemia is rare. The severity of the presentation of the child may be explained by the coincidence of these diseases.

Introduction

Down syndrome and β-thalassemia are prevalent genetic diseases worldwide.[1] The most common abnormality of chromosomal number in live-born infants is Down syndrome. Its incidence in live births is approximately 1 in 733.[2,3] Down syndrome is associated with congenital malformations and characteristic features.[3] Affected individuals are more susceptible to congenital heart defects (50%) such as atrial septal defects, ventricular septal defects, patent ductus arteriosus, and tetralogy fallot.[4-6] Down syndrome is also associated with an increased

risk of congenital and acquired gastrointestinal abnormalities (Celiac disease) and hypothyroidism.[7] The characteristic facial appearance with brachycephaly, flat occiput, hypoplastic midface, flat nasal bridge, unsplanting palpebral fissures, epicanthal folds, and large protruding tongue are often evident at birth.[8] Newborns also have short broad hands often with a single transverse palmar crease and a large gap between the first and second toes.[8] Hypotonia may lead to feeding problems and decreased activity.[9,10] Developmental delay is universal. The life expectancy for children with Down syndrome is approximately 50-55 years.[10,11] In about 95% of the cases, there are 3 copies of chromosome 21. Approximately one percent of the persons with trisomy 21 are mosaics while 4% have a translocation that involves chromosome 21.[8,12] Fusions at the centromere between chromosomes 13, 14, 15, 21, and 22, known as robertsonian translocation comprise the majority of translocation in Down syndrome. Chromosome analysis should be performed in every person suspected of having Down syndrome.[12]

Reduced or absent levels of alpha or beta chain of the hemoglobin characterize a heterogeneous group of hemoglobinopathies named Thalassemias.[3] β-thalassemia includes a group of hereditary hematological disorders characterized by deficiency or absence of synthesis of the β chains of hemoglobin, leading to variable phenotypes, ranging from severe anemia to clinically asymptomatic indi- viduals. People with β-thalassemia major commonly present with severe anemia within the first 2 years of life and need regular red blood cell transfusions.[13]

Case Report

Here, we report a 12-year-old boy with Down syndrome born to consanguineous parents and a 33-year-old mother. He was treated with regular transfusions every 20 days from 6 months due to anemia diagnosed with β-thalassemia major. The patient had a history of heart surgery at eighteen months old due to patent ductus arteriosus (PDA). He had a speech delay and examination revealed acanthosis nigricans on his neck area. His weight, height, and BMI were measured as 32 Kg, 129 cm, and 20, respectively. According to the genetic tests, his karyotype was 47, xy, +21.

Discussion

Down syndrome (Trisomy 21), the most common chromosomal abnormality, is commonly associated with a transient myeloproliferative disorder (TMD) and leukemias.[3] This syndrome leads to a collection of clinical features. It is commonly characterized by short stature, muscle hypotonia, atlantoaxial instability, reduced neuronal density, cerebellar hypoplasia, intellectual disability, and congenital heart defects (particularly atrioventricular septal defects).[8] Hypothyroidism, autoimmune diseases, obstructive sleep apnoea, epilepsy, hearing and vision problems, hematological disorders (including leukemia), recurrent infections, anxiety disorders, and early-onset Alzheimer's disease are health conditions individuals with Down syndrome are more susceptible to.[8] The coincidence of this syndrome with β-thalassemia major is uncommon. There are limited cases in the literature of the coincidence of Down syndrome and beta-thalassemia major.[12,14] In areas of high prevalence of β-thalassemia heterozygotes, the coincidence of the mentioned condition with another congenital disorder may be common.

Conclusion

The association between Down syndrome and β-thalassemia major is rare. The coincidence of β-thalassemia major and Down syndrome may explain the severity of the presentation of the child.[12] Down syndrome has also been described in association with hemoglobin S/b-thalassemia. Since the Thalassemia trait also remains a possible etiology for anemia in Down syndrome patients[15], it is suggested that Down syndrome should be checked for hemoglopathy in case of anemia.

Conflict of Interest

Authors have no conflict of interest.

Acknowledgments

The authors would like to thank the patient's family for their cooperation in this study.

Funding

Authors state no funding involved.

Ethical Considerations

The present study was approved by the Ethics Committee of Shahid Sadoughi University of Medical Sciences (IR.SSU.REC.1402.086).

Author's Contribution

NA.M. performed the examination of the patient, and was a major contributor in writing the manuscript. S.M. wrote and edited the manuscript. Both authors read and approved the final manuscript.

How to Cite: Mirhosseini NA, Mirhosseini S. A Case Report of Down Syndrome and β-Thalassemia Major Coincidence. World J Peri & Neonatol 2023; 6(1): 61-3.
DOI: 10.18502/wjpn.v6i1.14254

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