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Next generation sequencing revealed a rare case of co-existence of hereditary spherocytosis and dubin-johnson syndrome in a chinese gril: A case report

Yuan Li, Yang Li, Yang Yang, Wen-Rui Yang, Jian-Ping Li, Guang-Xin Peng, Lin Song, Hui-Hui Fan, Lei Ye, You-Zhen Xiong, Zhi-Jie Wu, Kang Zhou, Xin Zhao, Li-Ping Jing, Feng-Kui Zhang, Li Zhang

Abstract

BACKGROUND

Hereditary spherocytosis (HS) is a hereditary disease of hemolytic anemia that occurs due to the erythrocyte membrane defects. The Dubin-Johnson syndrome (DJS), which commonly results in jaundice, is a benign hereditary disorder of bilirubin clearance that occurs only rarely. The co-occurrence of HS and DJS is extremely rare. We recently diagnosed and treated one case of co-occurring HS and DJS.

CASE SUMMARY

A 21-year-old female patient presented to our department because of severe jaundice, severe splenomegaly, and mild anemia since birth. We eventually confirmed the diagnosis of co-occurring DJS and HS by next generation sequencing (NGS). The treatment of ursodeoxycholic acid in combination with phenobarbital successfully increased hemoglobin and reduced total bilirubin, direct bilirubin to 29.9 μmol/L.

CONCLUSION

The routine application of NGS can efficiently render a definite diagnosis when inherited disorders are suspected.

INTRODUCTION

Hereditary spherocytosis (HS) is a hereditary disease of hemolytic anemia that occurs due to the erythrocyte membrane defects caused by the gene mutation

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