IMAGE OF INTEREST

Korean J Intern Med 2021;36:232-233
https://doi.org/10.3904/kjim.2020.166

Renal coloboma syndrome with epilepsy

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An 8-year-old boy was brought to the nephrology and rheumatology department with a 3-day history of abnormal renal function and three episodes of twitching. Laboratory studies showed a creatinine level of 331.8 μmol/L, and color Doppler ultrasonography showed that the bilateral kidneys were small and unstructured (Fig. 1A and 1B). In addition, we found that the proband had an abnormal "morning glory" disc (Fig. 1C and 1D), and ultrasound showed a limited dent in each optic disc (Fig. 1E and 1F). These symptoms are similar to those found in renal coloboma syndrome. Interestingly, a 24-hour dynamic electroencephalogram revealed the occurrence of a paroxysmal epileptic wave during wakefulness (Fig. 1G). Such cases of renal coloboma syndrome with epilepsy were sporadic.

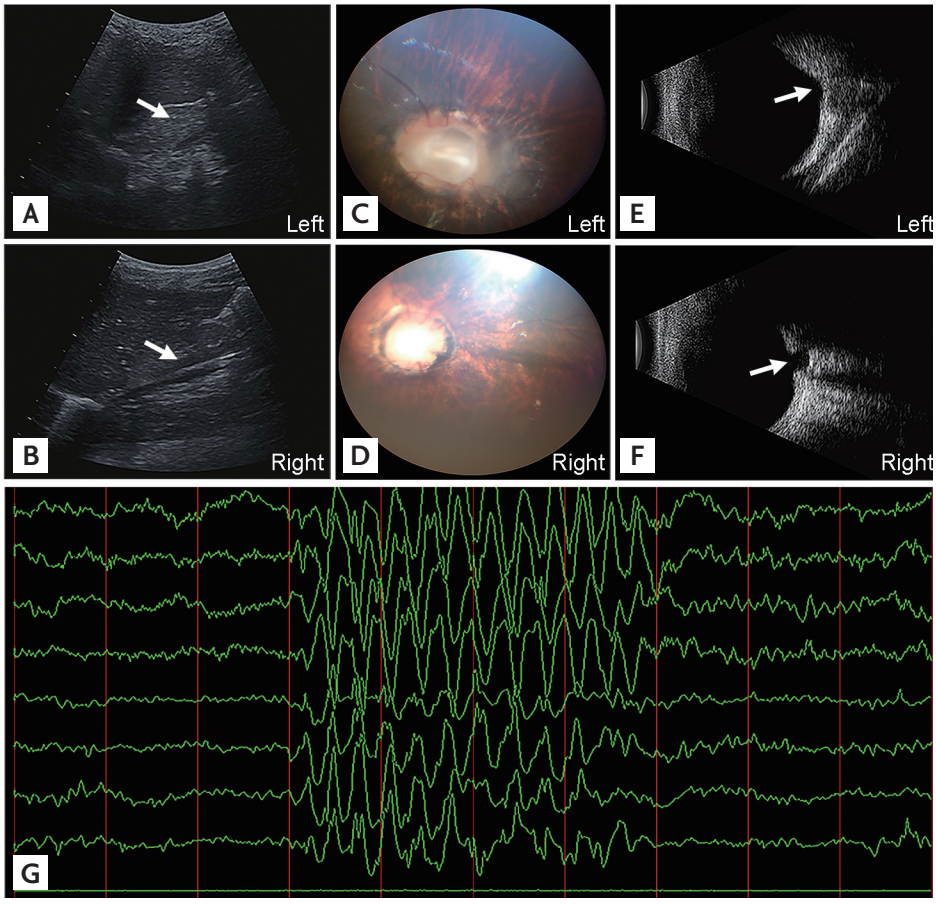

Figure 1. (A, B) Small bilateral kidneys (arrows). (C, D) Morning glory disc anomaly. (E, F) Optic disc coloboma (arrows). (G) Paroxysmal epileptic wave on a 24-hour dynamic electroencephalogram.

Received: April 21, 2020
Revised : May 6, 2020
Accepted: May 9, 2020

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pISSN 1226-3303
eISSN 2005-6648
http://www.kjim.org

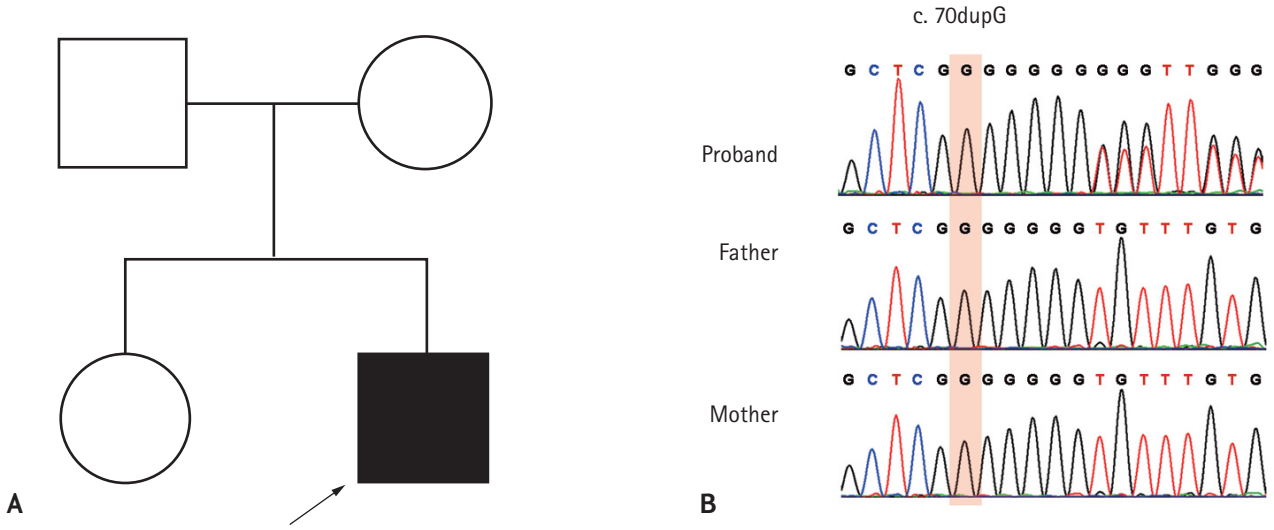

Figure 2. (A) Pedigree of family. (B) Identification of a *de novo* heterozygous paired box 2 (*PAX2*) mutation in the patient.

The patient had no family history (Fig. 2A) of similar diseases and had a c.70dupG (NM_003987) *de novo* heterozygous mutation in paired box 2 (*PAX2*) (Fig. 2B), which is responsible for renal coloboma syndrome. He was discharged when the tic subsided, and renal function improved slightly after 14 days of treatment with alkali therapy for acidosis and orally administered sustained-release sodium valproate tablets. A follow-up review was performed after 1 month, during which time the patient had been treated according to the doctor's advice at home. The patient showed maintenance of renal function along with a decrease in convulsions, but there was no significant improvement in his ocular disease.

Conflict of interest

No potential conflict of interest relevant to this article was reported.

Acknowledgments

We thank Xiantao Sun and Dan Wang for their discussion. This work was supported by a grant from the Research Project of the Science and Technology Department of Henan Province (Grant No. 192102310074).