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Case Presentation: We herein report a case of atypical Gillespie syndrome associated with a PAX6 mutation.

ABSTRACT

Background: Gillespie syndrome is a rare genetic condition characterized by mental retardation, cerebellar ataxia, and ocular anomalies, including microphthalmia, aniridia, and hydrophthalmos. We herein report a case of atypical Gillespie syndrome associated with PAX6 mutation.

Methods: We performed molecular genetic analysis on the PAX6 gene in our patient. No direct involvement of several known aniridia-associated mutations was found.

Results: The computer program 'fruit fly' (www.fruitfly.org/seq_tools/splice.html) predicts that this change will destroy the function of the ABR protein and is of unknown clinical significance. However, the computer program 'fruit fly' also predicts that this change will destroy the function of the ABR protein and is of unknown clinical significance. However, the computer program 'fruit fly' also predicts that this change will destroy the function of the ABR protein and is of unknown clinical significance. However, the computer program 'fruit fly' also predicts that this change will destroy the function of the ABR protein and is of unknown clinical significance.

Conclusions: Our patient's phenotype is consistent with a novel PAX6 mutation and is of unknown clinical significance. Further studies are needed to determine the clinical significance of this mutation.

REFERENCES