

Optic Pathway tumor in a novel KIAA1109 variant

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Abstract

Alkuraya-Kucinkas syndrome is a neurodevelopmental disorder due to mutation in KIAA1109 gene. We report a case of optic pathway tumor in a child with novel variant of KIAA1109. A plausible mechanism of gliomagenesis is considered based on influence of KIAA1109 on regulation of phosphatidyl inositol phosphorylation.

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