Mutations in the DHCR7 gene can lead to a rare genetic disorder known as Smith-Lemli-Opitz syndrome (SLOS). Individuals with SLOS have impaired DHCR7 activity, resulting in decreased cholesterol production and a range of developmental and physical abnormalities.

Aslan et al. aimed to attract attention to any possibility of a relation between SLOS and intracranial tumor development via a pediatric case with both intracranial high-grade neuroglial tumors and SLOS, and thus to contribute additional data to the literature on the togetherness of these two clinical conditions.

Method: In our clinic, we experienced an interesting case of a 10-year-old child with both SLOS and a huge brain tumor as rarely seen. Here, we reviewed the features and pathophysiology of SLOS and brain tumors via this case.

Results: The patient was operated on in our clinic, after, his brain tumor had been diagnosed, and his histopathology resulted in an undifferentiated malignant neuroglial WHO grade 4 tumor.

Conclusion: According to current literature, our case is the first report on the coexistence of SLOS and intracranial undifferentiated malignant neuroglial tumor. Common pathways like impaired sonic hedgehog (Shh) signaling pathways may be considered for the pathogenesis of a probable link between SLOS and brain tumors in further studies [1].

1)
Aslan A, Borcek AO, Pamukcuoglu S, Baykaner MK. Intracranial undifferentiated malign neuroglial tumor in Smith-Lemli-Opitz syndrome: A theory of a possible predisposing factor for primary brain tumors via a case report. Childs Nerv Syst. 2017 Jan;33(1):171-177. doi: 10.1007/s00381-016-3214-z. Epub 2016 Aug 15. PMID: 27526097.