Alagille Syndrome

The ocular signs of Alagille syndrome (ALGS) are diverse and can significantly aid in the diagnosis of the condition. The most common ocular finding is posterior embryotoxon, which is present in 78%-95% of patients. This is characterized by a prominent Schwalbe's line visible on slit-lamp examination.[1-3] Other notable ocular signs include optic disc anomalies, such as optic disc drusen and pseudopapilledema, which are found in a significant proportion of patients. Peripheral retinal abnormalities are also frequent, with chorioretinal changes being identified in up to 96% of patients. Additionally, iris abnormalities and diffuse fundus hypopigmentation have been reported in 45% and 57% of patients, respectively.[4] Less common but clinically significant findings include papilledema associated with intracranial hypertension, which may occur in a subset of patients, particularly post-liver transplantation. Microcornea and speckling of the retinal pigment epithelium are also observed in some cases.[4] These ocular manifestations, particularly posterior embryotoxon and optic disc anomalies, are critical for the early diagnosis and management of Alagille syndrome, as emphasized by the GeneReviews guidelines.[2]
https://pmc.ncbi.nlm.nih.gov/articles/PMC8254011/pdf/iovs-62-7-27.pdf

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