Prevalence of Cerebrotendinous Xanthomatosis (CTX) Among Patients Diagnosed With Juvenile-Onset Idiopathic Bilateral Cataracts

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Abstract (235/250 words)

Introduction: CTX, a rare bile acid synthesis disorder manifesting diverse signs and symptoms including childhood-onset bilateral cataracts, is often diagnosed and treated years after symptom onset, increasing risk of irreversible neurologic damage.[1] Caused by mutations in *CYP27A1*, CTX produces elevated plasma cholestanol (PC) and urinary bile alcohols (UBA). Metabolic testing for CTX among children with idiopathic acquired bilateral cataracts may aid earlier diagnosis and treatment of CTX.

Methods: The primary objective of this observational study was to evaluate CTX prevalence in patients aged 2 to 21 years at idiopathic bilateral cataracts diagnosis. Patients with PC levels ≥0.4 mg/dL or positive UBA prompted *CYP27A1* genetic testing at a CLIA-certified laboratory. The secondary objective was to assess other manifestations of CTX in patients with bilateral cataracts.

Results: 426 of 442 enrolled patients with a median age of 9.8 years (range, 1 month to 52.6 years) had available PC or urine samples, 28 (6.3%) met genetic testing criteria, and 4 (0.9%) tested positive for CTX. PC was 1.6-3.3 mg/dL in 4 patients who tested positive vs 0.38-0.65 mg/dL for patients who tested negative. 274 patients experienced at least 1 CTX-related symptom other than cataracts. The most common were eye disorders (29.4%), developmental delay (23.1%), and learning disability (21.3%).

Conclusion: CTX prevalence in idiopathic bilateral cataract patients was higher than population estimates (3-5/100,000).[2] Ophthalmologists can help diagnose CTX earlier by conducting metabolic testing in young patients with bilateral cataracts.

REFERENCES

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DISCLOSURES

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