

Diagnostic yield of workups ordered by pediatric ophthalmologists for bilateral pediatric cataracts at a tertiary pediatric hospital in the United States



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BACKGROUND

Children with unexplained bilateral cataracts routinely undergo testing for genetic, infectious, and metabolic etiologies. We evaluated the diagnostic yield of various tests ordered by pediatric ophthalmologists to evaluate bilateral cataracts in children at a single institution.

METHODS

We retrospectively identified all children with bilateral unexplained cataracts who underwent cataract surgery by a pediatric ophthalmologist at Children's Hospital Colorado from 2006 to 2022. We reviewed the results of genetic, infectious, and metabolic testing ordered by pediatric ophthalmologists to evaluate the cataracts in these children.

RESULTS

A total of 43 children met inclusion criteria. Of these, 34 (79%) had genetic testing, 34 (79%) had infectious disease testing, 33 (77%) had galactosemia testing, and 17 (40%) had urine-reducing substances testing performed during their cataract evaluation. Of the genetic tests ordered, 17 (50%) revealed a pathogenic mutation associated with cataracts. Twenty-three (68%) patients were IgG-positive for a TORCH infection, but no child was found to be positive on confirmatory testing. Of the galactosemia and URS tests ordered, 3 tests (9%) and 1 (6%) test were initially found to be abnormal, respectively, but confirmatory testing and clinical judgment ruled out metabolic disease in each case.

CONCLUSIONS

Genetic testing should be strongly considered in all cases of unexplained bilateral pediatric cataracts. Metabolic and infectious testing is best considered only after consultation with the child's pediatrician, guided by the patient's clinical context and the availability of genetic testing. (J AAPOS 2023;27:271.e1-5)
