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Abstract: Purpose: The purpose of this study was to determine whether the high rates of ocular problems described in previous retrospective reports of individuals with fragile X syndrome were present in a prospective sample of young boys. Fragile X syndrome is currently considered the leading hereditary cause of mental retardation, with prevalence estimates of 1:2500 to 1:5000 males.

Methods: Forty-eight boys with fragile X syndrome between the ages of 2.5 and 11 years were evaluated for ocular abnormalities. They received complete ophthalmic evaluations including assessment of visual acuity, cycloplegic refraction, ocular motility assessment, and dilated fundus examination.

Results: Approximately 25% of the children had clinically significant

ocular findings that included refractive errors (17%, primarily hyperopia and astigmatism) and strabismus (8%). Of the 42 children with quantifiable visual acuities, only 1 child, with diagnoses of nystagmus and hyperopia, had a Snellen visual acuity that was not within normal limits for his age. Three of the 5 children with gross measures of visual acuity had clinically significant findings: 2 had hyperopia greater than 3.5 diopters and 1 had esotropia. The other 2 children with gross measures of acuity and the 1 child without visual acuity assessment had cycloplegic refractions of +1.25 to +1.5 and ocular motility appeared normal.

Conclusions: These results suggest that previous reports of high rates of vision problems, particularly strabismus, in boys with fragile X syndrome may have resulted from selection bias. Although we did observe a higher prevalence of strabismus than that found in the general population (8% vs 0.5% to 1%), the proportion of children having strabismus in our sample was much smaller than that reported in other studies of children with fragile X syndrome (30% to 40%). However, 17% of the sample did have significant refractive errors. In addition to evaluating the ocular motility of children with fragile X syndrome, cycloplegic refraction should also be performed to determine whether refractive problems are present.

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