Autism spectrum disorder in visually impaired young children

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This commentary is on the article by Parr et al. on pages 917–921 of this issue.

Parr et al. report on a study of autism and autistic-like behaviour in children with optic nerve hypoplasia and/or septo-optic dysplasia.[1] The paper mainly addresses two important, and still controversial topics: (1) autism in visually impaired children; and (2) methods for assessing development and autism in visually impaired children.

The most commonly reported developmental disorder in blind children is autism. The possible factors accounting for this have been discussed over the years, ever since blindness and autism was first described by Keeler in 1958. Cass discussed the strong evidence that neurological disorders play a major part in deviant development and autism and stated: 'A phenomenon with such a remarkably high prevalence among the visually impaired population might be expected to arouse a considerable concern among parents, educators, therapists and doctors, and yet it has received relatively little attention in the literature' (p 118).[2]

Optic nerve hypoplasia is now a leading cause of vision loss in young children, a six-fold increase has been reported since 1977 and is now estimated at 10.9 per 100 000.[3] Several authors have reported high frequencies of autism and developmental disorders in children with optic nerve hypoplasia. Tornqvist et al.[4] reported additional impairments in 63% of a Swedish population. Our group[5] reported six children with autism and three with an autistic-like condition, with high frequencies of hormonal deficiencies and midline malformations (shown on computed tomography or magnetic resonance imaging), in a group of 13 blind children. In a prospective study focusing on neuroradiographic, endocrinological, and ophthalmic correlates of adverse developmental outcome, developmental delay was found in 71% of a population of 73 children.[3] Hypoplasia of the corpus callosum was highly correlated with deficiencies in the areas of personal, adaptive, communication, and cognitive functioning as well as with the overall development of the children. In cases of hypothyroidism, 93% had overall delay at the age of 5.[3]

The study by Parr et al. describes a large population (n=83) with severe or profound visual impairment. This is very unusual in studies of a specific ophthalmologic group like this; normally we have to be satisfied with much smaller groups.

The main weakness, however, is the retrospective design of the study. Over the study period (1977–2009) our knowledge of developmental disorders in visually impaired children and the methods for assessing it have changed. The retrospective study of case notes, estimating behaviour disorders and cognitive performance during one clinic visit, is questionable. These methods most certainly underestimate the true rate of autism spectrum disorders. Furthermore, it is not stated how many children in the study were blind (no light perception). Blindness in combination with brain disorder poses a definite risk of autism.

Intelligence testing in blind children is difficult and the problem has received extensive study. Few modern, well-standardized tests for visually impaired children are available. As the panorama of visual impairment has changed over the years, with more children now being diagnosed as visually impaired owing to causes affecting the posterior visual pathways and the brain, these norms are not accurate. Instead, we need in-depth clinical assessments using a variety of methods. The test performance by itself should be only one measure of intellectual ability. Observations of behaviour as well as teachers' and parents' reports and interviews are equally important.

The assessment of autism in blind children is a delicate process. The most common methods for scoring autistic behaviour include several items dependant on vision. Furthermore, no agreement has been reached as to distinguishing blindisms (stereotype, repetitive behaviour commonly found in blind children) from autism. The Autism Diagnostic Interview[6] is a comprehensive interview with parents that is useful also in blind children.

Hypothyroidism and hypoplasia of the corpus callosum are reported to be associated with developmental delay and autism. We look forward to the next paper by Parr et al. reporting their findings on imaging and hormonal data in this study group.

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