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Systemic Conditions in Children with Optic Disc Excavations

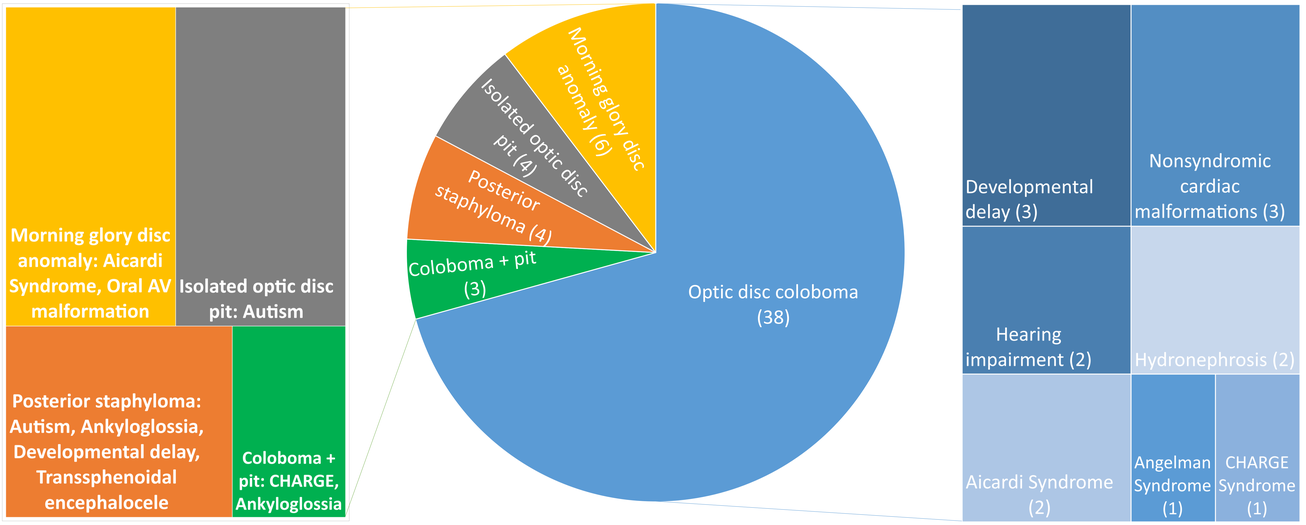
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Introduction

Optic disc excavations are congenital abnormalities of the optic disc that can significantly impair visual function. Approximately 30% of children diagnosed with an optic disc excavation are reported to have an associated systemic condition or syndrome. Here we report the incidence of systemic conditions in children with optic disc excavations in a well-defined paediatric population and propose a framework for holistic assessment that should be considered in these patients.

Methods

All children presenting to the paediatric ophthalmology department in Northern Ireland between 1 January 2003 and 1 January 2021 who had been identified as having an optic disc excavation were included. In our search we included patients with optic disc coloboma (ODC), posterior peripapillary staphyloma, optic disc pit and morning glory disc (MGD) anomaly. We retrospectively reviewed the electronic care record (NIECR) of each patient for investigations and results.





Optic disc coloboma with pit and chorioretinal coloboma



Peripapillary posterior staphyloma

Results

We identified 55 eyes of 44 patients. Of the 55 eyes; 38 had an optic disc coloboma, three additional eyes had coloboma and a disc pit, four had isolated disc pits, three had posterior peripapillary staphylomata and six had morning glory disc anomaly.

24 patients (55%) had an extraocular diagnosis; 17 (39%) were important systemic associations of optic disc excavation. Three patients had developmental delay: two associated with coloboma and one with posterior staphyloma. Three had Aicardi syndrome: two with coloboma and one with morning glory disc anomaly. Three patients with coloboma had non-syndromic cardiac defects. Two children with coloboma had an isolated hearing impairment. Two children with coloboma had features of CHARGE syndrome including one with a coloboma and a disc pit. One child with coloboma had Angelman Syndrome. Coloboma was associated with genitourinary anomalies in seven cases: two had hydronephrosis, one had a dysplastic polycystic kidney, one had anorchia (congenital absence of a testis) and another had an inguinal hernia. Other associations included retro-orbital cysts, transsphenoidal encephalocele with pituitary insufficiency, Alice In Wonderland Syndrome and epilepsy. There were also a number of common childhood diagnoses including asthma in two cases (coloboma), autism in two cases (one with posterior staphyloma and one with an optic disc pit), a facial arteriovenous malformation in one patient (morning glory disc anomaly) and ankyloglossia ("tongue tie") in a further two patients (coloboma with a disc pit and bilateral posterior staphylomata).

Aicardi syndrome is an X-linked dominant condition characterised by partial or complete absence of the corpus callosum, infantile spasms and chorioretinal lacunae. Less universal features include hypotonia, microcephaly, microphthalmia, skeletal dysgenesis and spasticity of the limbs. CHARGE syndrome is a highly variable constellation of conditions including coloboma, choanal atresia, cleft lip or palate, ear abnormalities, oesophageal atresia or fistula, genitourinary abnormalities and cardiac malformations. Mutations in the CHD7 gene are identified in approximately 90% of cases. Angelman Syndrome is an inherited condition that is usually sporadic and demonstrates the phenomenon of genomic imprinting, i.e. there is a mutation or inactivation of the maternal UBE3A gene. It is characterised by neurological disorders including developmental delay, intellectual disability, ataxia, speech problems and epilepsy. Ankyloglossia, caused by a congenitally short sublingual frenulum, is observed in up to 10% of children with or without systemic illness. Alice In Wonderland Syndrome is a rare neurological condition whereby patients (usually children) experience multi-sensory distortions of colour, form, chronology and scale as described in Lewis Carroll's novel *Alice's Adventures in Wonderland*.

Recommendations

Investigation of children with optic disc excavations should be guided by symptoms and examination findings. Minimum assessment should include full ophthalmological, neurological, developmental and cardiovascular examinations. Any ophthalmological disorders such as refractive error, strabismus and media opacity should be aggressively managed to maximise visual outcomes. Timely paediatric referral to assess for potential systemic associations (39% in our population) is appropriate in patients with optic disc excavations. Magnetic resonance neuroimaging is particularly helpful in patients with developmental or neurological symptoms and signs. Genetic testing can be very useful in patients with optic disc excavations in whom a syndromic association is suspected. Patients with developmental delay or intellectual disability may require lifelong input from multiple specialist services and outcomes are usually better with early diagnosis and appropriate signposting. Imaging of the renal tract should be considered particularly in children with clinical or biochemical evidence of genitourinary problems.

Conclusion

39% of our sample of 44 patients had an important systemic diagnosis associated with their optic disc excavation. These included Angelman Syndrome, Aicardi Syndrome, CHARGE syndrome, developmental delay not otherwise specified, sensory disabilities, neurocranial abnormalities and congenital renal and cardiac disorders.

Minimum assessment of children with optic disc excavations include full ophthalmological, neurological and cardiovascular examination. Paediatric referral is indicated in all children with optic disc excavations to ensure prompt holistic assessment.

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