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Original scientific paper

REFRACTIVE ERRORS AND BRUSHFIELD SPOTS IN DOWN SYNDROME WITH AUTISM SPECTRUM DISORDER: A CASE REPORT

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Abstract

Down syndrome is the most common chromosomal aneuploidy and is associated with an increased risk of various medical conditions, among which ophthalmological disorders are particularly significant. Their high prevalence, especially when co-occurring with autism spectrum disorder (ASD), complicates clinical assessment and necessitates a coordinated multidisciplinary approach. This paper presents the case of a six-year-old female patient with Down syndrome and ASD, in whom myopic astigmatism was diagnosed and pronounced Brushfield spots were observed along the entire limbus. The findings emphasize the importance of timely diagnosis, individualized strategies for the correction of refractive errors, and consistent parental involvement, which in synergy contribute to optimizing visual function and improving quality of life.

Keywords: Down syndrome, autism spectrum disorder, Brushfield spots, refractive errors

Introduction

Down syndrome, also known as trisomy 21, represents the most common chromosomal aneuploidy and one of the most prevalent genetic conditions worldwide. It occurs as a result of the presence of an additional copy of chromosome 21, with an estimated prevalence ranging from 1 in 400 to 1 in 1500 live births (Antonarakis et al., 2004; Kazemi et al., 2016). It is a genetic condition for which no specific primary prevention currently exists, while the risk increases significantly with advancing maternal age (Antonarakis et al., 2004). The clinical presentation is characterized by a recognizable phenotype that includes craniofacial features, mild to moderate intellectual disability, and a higher prevalence of comorbidities, most commonly congenital heart defects, hypothyroidism, gastrointestinal disorders, and impairments of vision and hearing (Shapiro, 2003).

Visual impairments are highly prevalent and heterogeneous, including strabismus, refractive errors, amblyopia, cataract, nystagmus, keratoconus, and retinal changes, which justifies the need for early ophthalmological assessment and continuous follow-up (Adio & Wajuihian, 2012; Sun & Kraus, 2023; Haseeb et al., 2022). Additionally, reduced accommodative ability is frequently reported in children with Down syndrome, independent of the presence of refractive errors (Woodhouse et al., 1996). As a therapeutic intervention, bifocal spectacles have been shown to be superior to monofocal lenses, providing improved visual acuity and reduction of accommodative deficit (Nandakumar & Leat, 2010). Research further indicates that the use of bifocals is associated with faster progress in literacy development, confirming their broader clinical and functional significance (Nandakumar et al., 2011). These findings emphasize the importance of routine assessment of accommodative function as part of comprehensive ophthalmological care in this population.

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Autism spectrum disorder (ASD) is a neurodevelopmental condition characterized by heterogeneous clinical expression. Its prevalence is estimated at approximately 1 in 44 children, with a pronounced sex imbalance favoring males (ratio approximately 4:1) (CDC, 2022). Clinical features manifest through difficulties in social communication, restricted and repetitive patterns of behavior, and frequently pronounced sensory characteristics (Mutti et al., 2018). The etiology is multifactorial and involves a complex interaction of genetic and environmental factors; cumulative scientific evidence does not support a causal association between vaccination and the development of ASD. Within the domain of visual functioning in children with ASD, sensory hyperreactivity (e.g., hypersensitivity to bright light or specific visual stimuli) and socio-perceptual characteristics such as avoidance of eye contact and difficulties in maintaining gaze are commonly reported, which may affect social interaction and the decoding of facial expressions. At the same time, some of these children demonstrate relative strengths in visual memory and pattern recognition, alongside difficulties in processing complex visual scenes. These characteristics justify the use of visually mediated interventions—visual schedules, picture-based communication systems (e.g., PECS), and social stories—which facilitate communication, structure daily routines, and clarify expectations in children with limited verbal abilities. According to current research, despite the limited number of studies, there is a clear association between autism spectrum disorder and various forms of visual impairment, particularly those related to refractive errors and strabismus (Perna et al., 2023).

Refractive errors, including myopia (nearsightedness), hyperopia (farsightedness), and astigmatism (impaired refraction due to an irregular shape of the cornea or lens), occur both in children with Down syndrome and in those with autism spectrum disorder. Although these conditions are also present in the general population, their prevalence is significantly higher in these specific groups. The most common therapeutic intervention is optical correction with spectacles or contact lenses, with the aim of improving visual acuity and preventing secondary complications such as amblyopia.

Table 1. Visual characteristics in Down syndrome and autism spectrum disorder (ASD)

Characteristic	Down syndrome	ASD
Refractive errors	The most common refractive error is myopia (nearsightedness) (Surasmiati et al., 2025).	Approximately 48.4% of children present with refractive problems, most commonly hyperopia and astigmatism; myopia is more frequent in Asperger syndrome (≈18%) (Gutiérrez et al., 2022).
Strabismus	Esotropia (inward deviation of the eyes) is more common, with a higher prevalence than in typically developing children (Haugen & Hovding, 2001).	Present in more than 15% of patients. The most common form is exotropia (outward deviation of the eyes) (Gutiérrez et al., 2022).

What are Brushfield spots?

Brushfield spots are small whitish or yellowish formations located along the periphery of the iris, first described by the British physician Thomas Brushfield in 1924. They are observed in a

considerable proportion of individuals with Down syndrome, most commonly ranging from one-third to two-thirds of cases, with greater visibility in individuals with lighter-colored irises due to the lower concentration of melanin (Figure 1). Morphologically, they are arranged in a ring-like pattern along the outer margin of the iris, often more pronounced in the inferior segment, and their size may vary from subtle and barely noticeable to more prominent and clearly visible.

Figure 1. Child with Down syndrome and Brushfield spots

Although Brushfield spots are considered a benign ophthalmological finding without functional impact on vision, they have significant clinical value and have long been regarded as a characteristic phenotypic marker in individuals with trisomy 21. In the contemporary context, they do not represent a diagnostic criterion on their own, as confirmation of Down syndrome is based on genetic analysis. Nevertheless, their presence still retains indicative significance and may serve as a useful visual indicator during clinical assessment, particularly in settings where additional diagnostic resources are limited (da Cunha & Moreira, 1996).

Case report

General patient information: A six-year-old female patient from the village of Shemshevo (Tetovo) with a confirmed diagnosis of Down syndrome and autism spectrum disorder (ASD).

Anamnesis and perinatal data: The mother's pregnancy was uneventful, with no recorded complications; delivery was performed by cesarean section.

Reason for referral: The patient was brought for an ophthalmological examination due to symptoms of photophobia, manifested by squinting even in minimal light conditions.

Clinical course: Due to cooperation difficulties associated with ASD, four separate visits were required to establish rapport and successfully perform the ophthalmological examination.

Findings of the ophthalmological examination: Refraction assessment and biometry revealed a visual acuity of 0.8 in the right eye and 0.7 in the left eye. Myopic astigmatism was identified with the following values:

OD: −0,75 −1,25 ax 20°

OS: −1,00 −1,00 ax 170°

During the objective examination, the presence of Brushfield spots was observed, distributed along the entire limbal circumference.

Therapy and recommendations: The patient was prescribed corrective spectacles with the appropriate refractive correction and ultraviolet (UV) protection, along with the recommendation to wear a hat when exposed to sunlight.

Follow-up and outcome: At the follow-up examination after one month, the parents reported that the child wears the spectacles regularly and without resistance, including during outdoor activities. Successful adaptation to the corrective therapy was observed.

Discussion

The presented case illustrates several important aspects of clinical practice when working with children with Down syndrome in combination with autism spectrum disorder. First, the difficulties in establishing cooperation during the ophthalmological examination confirm the complexity of these conditions. In children with autism, a more flexible approach, multiple visits, and adapted examination methods are often required, which is consistent with previous research indicating that sensory hypersensitivity and difficulties in social interaction may complicate medical management (Mutti et al., 2018).

Second, the diagnosed myopic astigmatism is consistent with the well-documented high prevalence of refractive errors in individuals with Down syndrome. Haugen and Hovding (2001) and Surasmiati et al. (2025) report that myopia and astigmatism are among the most common visual disorders in this population, with a significantly higher prevalence compared to the general pediatric population. This highlights the importance of early and regular ophthalmological assessment in order to ensure timely detection and correction of these conditions, thereby reducing the risk of amblyopia and enabling optimal visual development.

Third, the presence of Brushfield spots, although benign, has diagnostic significance. As reported by da Cunha and Moreira (1996), they occur in 35–78% of individuals with Down syndrome and are more evident in individuals with lighter-colored irises. In this case, the spots were pronounced and evenly distributed along the limbus, confirming their role as a phenotypic characteristic. Although genetic testing currently represents the gold standard for the diagnosis of trisomy 21, these spots still retain clinical relevance as an indicative marker.

Finally, the successful adaptation to the prescribed spectacle correction and the regular use of sun protection emphasize the importance of parental involvement in the treatment process. Family support and consistency are key factors in achieving positive outcomes, particularly in children with complex comorbidities such as Down syndrome and autism spectrum disorder.

Conclusion

The presented case illustrates the importance of early ophthalmological evaluation and an individualized approach in children with Down syndrome and autism spectrum disorder. The presence of refractive errors, such as myopic astigmatism, together with the characteristic phenotypic finding of Brushfield spots, indicates the need for regular ophthalmological screening in this population. Timely correction with appropriate optical aids, combined with education and active parental involvement, contributes to the improvement of visual function and facilitates the patient’s daily functioning.This case further confirms the value of a multidisciplinary approach that integrates ophthalmological, pediatric, and psychological care, thereby ensuring comprehensive and holistic management. Such an approach is essential for optimizing quality of life and improving long-term outcomes in children with complex developmental and medical needs.

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