# Decoding optic nerve:
## How OCT reveals the secrets of congenital eye disorders

*On December 6, 2024, Athanasia Skriapa-Manta defended her thesis "Congenital optic disc malformations: prevalence, clinical and OCT findings, longterm follow-up and quality of life" at the Dept. of Clinical Neuroscience, Karolinska Institutet. Her main supervisor was Adjunct Prof. Kristina Teär Fahnehjelm, Karolinska Institutet. Dept. of Clinical Neuroscience, Division of Eye and Vision. Co-supervisors were Adjunct Prof. Ronny Wickström, Dept. of Women's and Children's Health, Division of Neuropediatric Unit, and Associate Prof. Britt Marie Anderlid, Dept. of Molecular Medicine and Surgery, both at Division of Neuropediatric Unit, Karolinska Institutet.*

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Congenital optic disc anomalies, also known as malformations, are structural abnormalities caused by abnormal development of the optic nerve during early prenatal life, typically in the first trimester. These anomalies can affect one or both eyes and may occur in isolation, in association with systemic conditions, or as part of a genetic syndrome. Visual outcomes can range from minimal to severe vision loss.

The most common congenital optic disc anomalies include:

- **Optic nerve hypoplasia (ONH):** An underdeveloped optic nerve with reduced nerve fibers.
- **Optic nerve aplasia:** A rare condition where the optic nerve is entirely absent.
- **Optic disc coloboma (ODC):** A large, excavated defect of the optic nerve, often extending into the retina.
- **Morning glory disc anomaly (MGDA):** A funnel-shaped optic disc resembling a morning glory flower.
- **Optic nerve pit:** A small depression or hole in the optic disc.
- **Optic disc staphyloma:** Thinning and protrusion of the posterior sclera into the orbit.

These rare and often subtle conditions can be difficult to diagnose, and only limited studies have been published. This thesis aimed to contribute to the field by using advanced imaging tools, particularly optical coherence tomography (OCT).

In our first study, we found the prevalence of ODC in the Swedish pediatric population to be 8.9 per 100,000, making it the second most common anomaly after ONH. Visual outcomes varied, but were generally favorable when the fovea was unaffected. In cases of isolated ODC, pediatric referral was typically unnecessary if routine health checks were normal.

Our second and third studies focused on the role of OCT in diagnosing ONH. Traditionally, diagnosis is clinical, using ophthalmoscopy and fundus photography. However, OCT offers objective measurements of retinal layers. In one study of 37 ONH eyes from 20 patients, we found that visual acuity correlated with specific OCT parameters, such as the peripapillary retinal nerve fiber layer (pRNFL), ganglion cell complex (GCC), and Bruch's membrane opening (BMO). Thinning in these layers often indicated visual field defects and helped predict their location.

In a larger cohort of 52 ONH eyes and 17 fellow eyes with high-quality OCT images, we used a semi-automated analysis method in MATLAB. This approach allowed detailed analysis of optic disc and macular structure. While OCT proved valuable in characterizing ONH, we concluded that the relationship between retinal structure and visual function is complex and multifactorial, requiring individualized assessment.

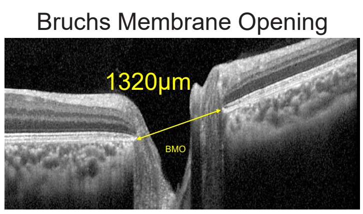


*Figure 1. Measurement of Bruchs Membrane Opening*

**Key points:**

- Evaluating both the optic disc and macula is important to understand visual function in early childhood. Parameters such as BMO and GCC thickness show strong correlations with visual acuity and patterns of visual field defects.
- These results support a more individualized approach to the diagnosis, treatment, and long-term management for children and adolescents with ONH, promoting personalized rehabilitation and follow-up strategies.

In our fourth study, we conducted a long-term follow-up of 31 patients with ONH. We observed that visual acuity remained stable over time, although a trend toward myopic shift was noted. Quality-of-life (QoL) scores were generally low, correlating with visual acuity. However, many ONH patients also experienced hormonal, neurological, or behavioral issues, which likely contributed significantly to reduced quality of life.

***Future directions:***

- Congenital optic disc malformations are rare, and the populations included in different studies are small. There is a need for more structured data collection in larger cohorts of patients to expand our knowledge regarding OCT findings, long-term follow-up, and health-related QoL. Using newer, more advanced OCT devices with faster acquisition times and the ability to image the entire retina in one scan could offer new insights.

***References***

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