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Congenital **Aniridia**

Exploring Visual Disabling Manifestations in the Ocular Surface and Ocular Fundus through Clinical and Translational Approaches

Erlend Christoffer Sommer Landsend (M.D.) defended his doctoral thesis on February 13 this year at the Faculty of Medicine, University of Oslo. Congenital aniridia is a panocular disease, which is characterized by hypoplasia of the iris and the retinal fovea at birth. Foveal hypoplasia is the main cause of congenital reduced vision in aniridia. Most patients are born with low vision. Serious ocular complications may reduce the vision further in patients with aniridia. These include aniridia-associated keratopathy, which leads to progressive opacification of the cornea, pain, and often considerable visual impairment or blindness.

In his thesis, Landsend included 35 patients with aniridia and 21 healthy individuals. The participants were examined extensively regarding dry eye disease. Cytokine concentrations in the tear fluid were measured. Next, 14 of the 35 aniridia patients underwent autofluorescence imaging of the ocular fundus. The images were compared with 14 age- and gender-matched healthy individuals.

Landsend and colleagues detected more severe dry eye disease in aniridia patients than in healthy participants. Importantly, development of dry eye disease and keratopathy were interconnected. Aniridia patients showed more inflammation at the ocular surface in terms of increased levels of a number of pro-inflammatory tear cytokines. Higher cytokine concentrations were associated with more severe dry eye disease. Landsend found that autofluorescence imaging could be a useful tool in evaluation of foveal hypoplasia and give information that is not available through other clinical tools. Further, the findings give clues to the pathophysiology of foveal hypoplasia and to foveal development in general.