

# TIMOTHY A. JOHNSON MEDICAL SCHOLAR LECTURE SERIES

Presented by the Fralin Biomedical Research Institute at VTC and the Virginia Tech Carilion School of Medicine



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## *In Person Lecture: Albinism: Can It Become a Treatable Disease?*

Oculocutaneous albinism (OCA) is a genetically and phenotypically heterogeneous, autosomal recessive condition characterized by reduced melanin in the hair, skin and eyes. Difficulties with vision such as reduced best-corrected acuity (due to abnormal development of fovea, a specialized area of the neural retina) and difficulty with glare sensitivity (likely due to reduced absorption of stray light) are common in people with albinism. We do not understand why reduced melanin in the pigmented layers of the eye affect the development of the fovea and other aspects of the neural retina (where pigment genes are not expressed). If this process were understood, however, we may be able to devise treatments that improve vision if exacted during the period of foveal development soon after birth. This lecture will review what is known about the clinical presentation, genetics and biology of albinism, including discussion of the melanogenic pathways; discuss data on preclinical and pilot clinical trial repurposing of the FDA-approved drug, nitisinone (NTBC) and efforts for large-scale drug screens to activate tyrosinase, the first and rate-limited enzyme in melanin synthesis; and cover recent characterization of RPE cells from induced-pluripotent stem cells (iPSCs) as a method for identifying differences between albinism and control cells with a focus on druggable targets.

**FRIDAY, SEPT. 16 at 1 p.m.**

Room M203, 2 Riverside Circle.

Watch live via Zoom at <https://virginiatech.zoom.us/j/82257173281> or at <https://fbri.vtc.vt.edu/events/live-webcast.html>.

