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Commentary Open Access

Commentary: Effect of Agalsidase Beta on Corneal Verticillata in Patients with Fabry Disease

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Commentary

In our previous article entitled, "Ocular manifestations of Fabry disease within in single kindred", we describe the ocular and systemic manifestations of Fabry disease. Fabry disease is a lysosomal storage disease inherited in an X-linked fashion which results in deficient enzyme levels of alpha-galactosidase A (AGAL), an enzyme important for the breakdown of various glycoproteins in the body. AGAL deficiency prevents the effective metabolism of the glycoprotein globotriaosylceramide (Gb3). Consequently, Gb3 deposits within lysosomes in virtually every tissue in the body including the eyes, kidneys, brain, and heart. Increasing storage of Gb3 and the resultant intrinsic inflammatory reactions lead to increased disease burden in the patient with disease progression. The initial symptoms can include angiokeratomas (telangiectatic cutaneous lesions), acroparesthesia (severe pain in hands and feet), hypohidrosis (inability to sweat), gastrointestinal dysmotility, and various ocular manifestations [1].

The most commonly reported ocular finding is corneal verticillata, a bilateral, whorl-like pattern of cream-colored lines usually found in the inferior cornea. These lines are caused by the accumulation of Gb3 at the level of Bowman's membrane, which radiate out in a vortex pattern. In a slit lamp examination, the degree of corneal verticillata can range from very faint to quite obvious but rarely affects visual acuity though glare has been reported.

Agalsidase beta is recombinant alpha-galactosidase A enzyme and has revolutionized the treatment of Fabry patients. While this enzyme has shown to alleviate symptoms and progression of Fabry disease, there has been no study to date evaluating the effect on corneal verticillata and whether or not resolution occurs [2].

We presented our data at the 2015 American Academy of Ophthalmology (AAO) Annual Meeting in which thirteen Fabry patients with documented corneal verticillata on examination and being treated at that time with enzyme replacement therapy (agalsidase beta) participated in our analysis. On two year follow-up, careful slit lamp examinations were performed to analyze the anterior segment for the presence or resolution of corneal verticillata. Three out of the thirteen patients (23%) had complete resolution of corneal verticillata in both eyes.

All evaluated Fabry patients were being treated with agalsidase beta and had corneal verticillata, which generally does not affect vision and is readily recognizable by slit lamp examination. Corneal verticillata in patients with Fabry disease appear to resolve in about a quarter of patients when treated with agalsidase beta.

These results provide objective clinical evidence of the efficacy of enzyme replacement therapy (ERT) in Fabry disease. Whether the resolution of corneal verticillata with ERT can be a clinical gauge to the patient's response to therapy is still under investigation and is a promising premise for future studies.

Overall, the eye care provider can play a crucial role in the early recognition of ocular manifestations of Fabry disease and decrease both the time to accurate diagnosis and the delay in the institution of disease-modifying therapy.

References

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