

## Late Onset Retinal Degeneration

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### INTRODUCTION

Late-onset Retinal Degeneration is a rare autosomal dominant retinal disorder. Symptoms are often related to challenges with light adaptation and progressive visual field loss [1]. Characterized by thickening of retinal pigment epithelium (RPE) due to lipid deposits (Drusens) RPE atrophy, choroidal neovascularization (CNV) and severe visual loss due to photoreceptor cell death with the progression towards severe central and peripheral degeneration [2]. With characteristics as such, it has significant resemblances to Age-related Macular Degeneration (AMD). The most common form of diagnostic modalities is the use of Optical coherence tomography angiography (OCT-A), however, others include Fluorescein angiography, Indocyanine green angiography and examination of the retina. Further progression of the disease can lead to severe CNV and/or glaucoma secondarily to LOR-D.

### DISCUSSION

LOR-D is known to be related to a missense mutation (S163R) in the C1QTNF5 gene that encodes C1q and Tumor Necrosis Factor Related Protein 5 (C1QTNF5) [1]. Thus, resulting in changes a highly conserved serine to arginine (Ser163Arg), however, not all patients presenting with LOR-D have C1QTNF5 mutation. It is also noted that Ser163Arg mutation appears to cause age-related accumulation of deposits such as Drusens between Bruch's membrane and RPE [3]. Furthermore, despite the normal role of C1QTNF5 remaining unclear; it is highly expressed within the eye in the retinal pigment epithelium (RPE), ciliary body and lens epithelial cells [4]. Long anterior Zonules (LAZ) are also often found within the lens of LOR-D patients [3]. However, the roles of these fibers are unknown and has been long theorized that LOR-D may create challenges during and post-operations. In contrast, in recent studies suggest that LAZ does not play a significant role in the stability of IOL, thus, cataract surgery secondary to LOR-D is reasonably a safe procedure provided that long term maintenance of IOL stability achieved [4].

### DIAGNOSING

With so much being unknown, diagnoses of LOR-D at early stages can be challenging due to symptom similarities to AMD. Often times vision abnormalities are noticed around midlife; 50-60 years of age for disease processes with Drusen deposits. It should also be noted that late-stage non-exudative AMD is similar to STGD1 due to being characterized by RPE atrophy at the posterior pole which presents another diagnose challenge [5]. With challenges as such, LOR-D diagnosis should include thorough evaluations of medical, family, social history, review of system, physical and ophthalmic examinations. With the use of OCT-A, these Drusen spot like deposits can be visualized throughout the retina. Recent studies of choroidal flow signal using OCT-A, suggest that there are definitive phenotypic differences between STGD1 and AMD, thus, implicating different role of the choroid in the pathogenesis of RPE atrophy in both diseases [5]. Therefore, treatment approaches between diseases could vary in effectiveness due to disease processes.

### REFERENCES

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