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# Nucleome Informatics launches DrSeq IRD test for comprehensive genetic diagnosis of Inherited Retinal Diseases

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**DrSeq IRD panel includes 850 genes and thousands of known and novel variants, providing superior diagnostic accuracy**

Nucleome Informatics, a Hyderabad-based genomics services provider, has announced the launch of its DrSeq IRD panel, a revolutionary genetic test for diagnosing Inherited Retinal Diseases (IRDs).

This test is among the most comprehensive available globally, designed to improve early detection and disease management, especially in individuals with a family history of blindness.

IRDs represent a genetically diverse spectrum of disorders that affect the retina, leading to vision impairment or blindness. In India, the prevalence of IRDs ranges from 1 in 350 to 1 in 2000, varying by region and factors such as consanguinity.

Accurate diagnosis of these conditions has been challenging due to their clinical and genetic heterogeneity, with over 300 genes associated with IRDs. However, most cases are caused by mutations in 20 key genes.

Nucleome's DrSeq IRD panel covers an extensive range of IRDs, including Leber Congenital Amaurosis (LCA); Cone Dystrophy; Cone Rod Dystrophy; Retinitis Pigmentosa (RP); Stargardt's Disease; Maclaren Dystrophy; Congenital Stationary Blindness; Vitelliform Macular Dystrophy; Bardet-Biedl Syndrome; Usher Syndrome; Choroideremia; Achromatopsia; and Bestrophinopathy

The DrSeq IRD panel includes 850 genes and thousands of known and novel variants, providing superior diagnostic accuracy. The test helps identify the genetic mutations responsible for specific IRD types, which is critical for tailoring treatment options, including gene therapy.

In collaboration with LV Prasad Eye Institute (LVPEI), DST (Department of Science and Technology) of the Government of India and Korean partners, including Seoul Eye Hospital, Nucleome Informatics has developed the DrSeq IRD panel by sequencing 300 IRD patients and 200 healthy family members from India.

One of the unique aspects of the DrSeq IRD panel is its saliva-based collection method, which eliminates the need for blood

samples.