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complement factor H as
a major risk factor for age-
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degeneration, vision
science is also at the
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Molecular Diagnosis and
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management in a successful ophthalmic genetics practice requires expertise found in ophthalmology, medical genetics, genetic counseling, clinical molecular genetics, and often pediatrics. The team must apply rapidly changing molecular advances in the recent literature to patient care.

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our research focuses on
inherited disorders of the
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instructions which
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families. Our aim is to increase understanding of how these mutations cause disease, how these diseases progress over a person's lifetime and the further impact of other biological and environmental factors.

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Contd. Molecular

genetic studies of the RB1
gene can now be

proposed to all patients

with familial or sporadic

unilateral or bilateral

retinoblastoma. Genetic

consultation in

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paediatric oncology and
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managing the child.
Family informed about
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Patient ' s pedigree
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team working between the Centre for Genomic Medicine and The Manchester Royal Eye Hospital. We see patients and families with conditions that affect vision and are thought to have a genetic or inherited basis.

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publication is to enhance
mutual understanding
and communication

between
ophthalmologists,
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biomedical researchers.

In the introductory

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chapter, current genetic paradigms and experimental genetic approaches relevant to the nature of hereditary disorders are discussed.

Genetics in
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Stargardt disease
(STGD1; MIM 248200)
is the most prevalent
inherited macular

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dystrophy and is associated with disease-causing sequence variants in the gene ABCA4 .

Significant advances have been made over the last 10 years in our understanding of both the clinical and molecular features of STGD1, and also the underlying pathophysiology, which has culminated in

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long understood that the organization of the visual pigment genes for human color vision would have to be complex enough to accommodate the production of 3 opsin types in 3 spectral classes of cone.

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