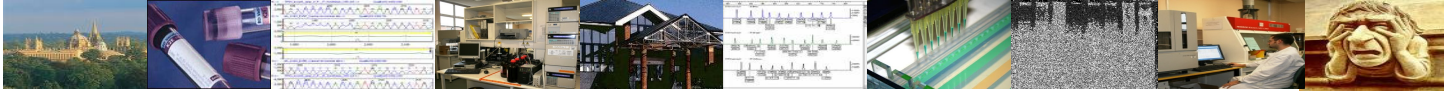


Oxford Molecular Genetics Laboratory



Stargardt disease type 1 (STGD1) / Fundus Flavimaculatus (FF) – OMIM 248200

INTRODUCTION

Stargardt disease type 1 (STGD1) is the most common autosomal recessive retinal dystrophy with an estimated prevalence and carrier rate of 1 in 10,000 and 1/33-1/50, respectively. Characteristic features include significant loss in central vision in childhood to early adulthood, progressive bilateral atrophy of the photoreceptors and underlying retinal pigment epithelium and the presence of yellow-white flecks within the macula and in some cases in the mid-retinal periphery as well. The term fundus flavimaculatus (FFM) is sometimes used interchangeably with STGD1 but is usually used when the flecks are more widely distributed and scattered throughout the fundus. However, it is accepted that STGD1 and FFM are manifestations of the same disorder.

STGD1/FFM is caused by loss-of-function pathogenic mutations in the *ABCA4* gene (1p13-p22, 50 coding exons). The *ABCA4* gene is highly penetrant although intrafamilial variation has been reported. Pathogenic mutations in *ABCA4* are also responsible for some cases of other forms of retinal degeneration, including autosomal recessive cone-rod dystrophy (arCRD) and autosomal recessive retinitis pigmentosa (arRP).

TESTING AND REFERRALS

Diagnostic:

- Clinically affected patients referred by ophthalmic genetic specialists

Family tests:

- Confirmation of carrier status in parents of affected patients with two pathogenic mutations
- Testing of at-risk relatives of affected patients with two pathogenic mutations
- Referrals accepted from Clinical Genetics

STRATEGY AND TECHNICAL INFORMATION

- Mutation screening in diagnostic referrals is undertaken by fluorescent sequencing of the coding regions and intron/exon boundaries of the *ABCA4* gene. If two pathogenic mutations are not detected dosage multiplex ligation-dependent probe amplification (MLPA) analysis is performed to test for large scale rearrangements (to our knowledge there have been two large scale deletions reported in the literature).
- When a pathogenic mutation/s has been identified in an individual, subsequent carrier testing of parents and other relatives involves testing for the familial mutations only.

TARGET REPORTING TIMES AND COSTS*

Diagnostic:	8 weeks	£900
Family mutation test:	10 days	£221

*Referrals from outside of the Oxford Regional Molecular Genetics Laboratory contracted area.

* Non NHS patients are subject to a 10% surcharge and payment must be agreed prior to testing.

N.B. Details are correct for the date of printing only

Sample Requirements:

5-10ml venous blood in plastic EDTA bottles (>2ml from neonates)

A completed DNA request card should accompany all samples.

Consent for testing is assumed to have been obtained by the referring consultant.

Contact Details:

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Useful links:

<http://www.ukgtn.org/>
<http://www.eddnl.com/>
<http://www.geneclinics.org/>