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# MAC. Microphthalmia, Anophthalmia and Ocular Coloboma

NIHR BioResource – Rare Diseases study project

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## Summary

Ocular maldevelopment encompasses a clinical spectrum of rare genetic disorders linked to structural defects in early eye formation. It is responsible for more than one-third of blindness and severe visual impairment in children worldwide. It involves whole eye abnormalities (e.g. microphthalmia [abnormal small eye], anophthalmia [complete absence of the eye] and ocular coloboma [cleft of the eye], collectively known as MAC) and affects the anterior segment (e.g. corneal opacity, iridocorneal anomalies, aniridia and cataract).

The UK incidence of MAC is 10.4 per 100,000 live births. They can be found in isolation or coupled with other ocular conditions (complex) such as the aforementioned anterior segment anomalies and retinal dysplasia, and up to 60% of cases are syndromic. They are a group of genetically heterogeneous conditions with significant inter- and intra- familial phenotypic variability, but the overall genetic diagnostic rate is less than 10%. There are no treatments for these conditions, just supportive management, however much research is underway to identify novel disease-causing variants and potential therapeutic targets that may encourage post-natal eye growth and visual development.

The main aim of this project is to gain a better understanding of the genotype and phenotype in patients with MAC, for clinical risk stratification and to potentially identify genetic modifiers affecting to influence disease severity. To achieve these objectives, we will collect detailed phenotyping data of this group of disorders and blood samples to identify the underlying genetic basis for the patient's condition.

## Recruitment Criteria

### Inclusion

Patient with a clinical diagnosis of microphthalmia, anophthalmia and/or ocular coloboma with or without other ocular (complex) or systemic features (syndrome).

We encourage the recruitment of multiple affected members of a single family.

Parents of a patient with a clinical diagnosis of MAC.