**Call for collaborative research on rare eye diseases**

These calls for collaborative projects typically aim to rapidly build up consistent clinical series for rare monogenic disorders, in order to better delineate the clinical spectrum and natural history of disorders in the field of the ERN-EYE.

**Title: Association of ABCA4 macular dystrophy and NF1 – disease burden**

Targeted gene/disorder under study: ABCA4 Retinopathies, NF1

Abstract

ABCA4 Retinopathies are the most common cause of juvenile macular dystrophy. This is an autosomal recessive disease group, with a carrier frequency reported to be as high as 1 in 20 (depending on the population). Furthermore, NF1 is one of the most common autosomal dominant disorders, with estimated prevalence between 1 and 3,000. Both disorders have ophthalmologic implications. People with both ABCA4 Retinopathies and NF1 (3 individuals in Romania and 1 in literature) may have increased disease burden.

With this study we would like to answer the following questions:

In individuals with both ABCA4 Retinopathies and NF1

• What is the quality of life (general and vision specific)?

• What is the age range at which the visual acuity decreased?

• What are the associated comorbidities?

• Does ophthalmological surveillance in NF1 result in early detection of ABCA4 Retinopathies?

Coordinating clinician

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Specific requirements beyond clinical data and genotype data sharing:

1. Re-analysis of DNA samples: No
2. Re-contact patients –yes
3. Resampling of patients: No
4. Linked to a translational/basic research project: No