CNC
CENTER FOR NEUROSCIENCE AND CELL BIOLOGY
UNIVERSITY OF COIMBRA, PORTUGAL

Center for Neuroscience and Cell Biology

Neurogenetics and Vision Related Disorders Laboratory

University of Coimbra

Request Form for Genetic testing of Vision related disorders

Per patient, two EDTA blood 3 ml tubes should be taken and each tube should be labelled with surname, forename and date of birth.

Store blood at room temperature (do not freeze). This filled out request Form must accompany the samples.

Referring Clinician (person to whom result will be sent)

Forename: ________________________________ Surname: ___________________________

Hospital/Institute: ___________________________ Department: _________________________

Request Date: _________________ Signature:_____________ Email: _________________________

Patient Data

Forename: ________________________________ Surname: ____________________________

Date of birth: ___/___/___ Gender: ___________ Ref. Number ________________________

Invoice Address: ____

____

Clinical Information

Clinical Diagnosis: ____________________ known mutation? No ☐ Yes ☐ which?___________

Patient (*index* case) ☐ Patient (*familiar* case) ☐ Asymptomatic (*familiar* case)* ☐

* The Laboratory will not proceed with predictive testing without a copy of the signed informed consent and genetic counselling

Aditional Information (Please include any relevant clinical information such as symptoms, family history and pedigree if appropriate)

Tests Required (please tick the correct choice)

Retinitis pigmentosa

☐ All coding exons of Rhodopsin gene (*RHO*)

Nanophthalmia

☐ All coding exons of *MFRP* gene

Anophthalmia / Microphthalmia (A/M)

☐ All coding exons of *OTX2* gene
☐ All coding exons of *RAX* gene
☐ All coding exons of *SOX2* gene

☐ Other Test_________________

Test for known mutation

☐ Please give details of mutation _________________

and proband's name ___________________________

To be filled in by laboratory

Sample No. ________________________ Arrival Date: ___/___/___