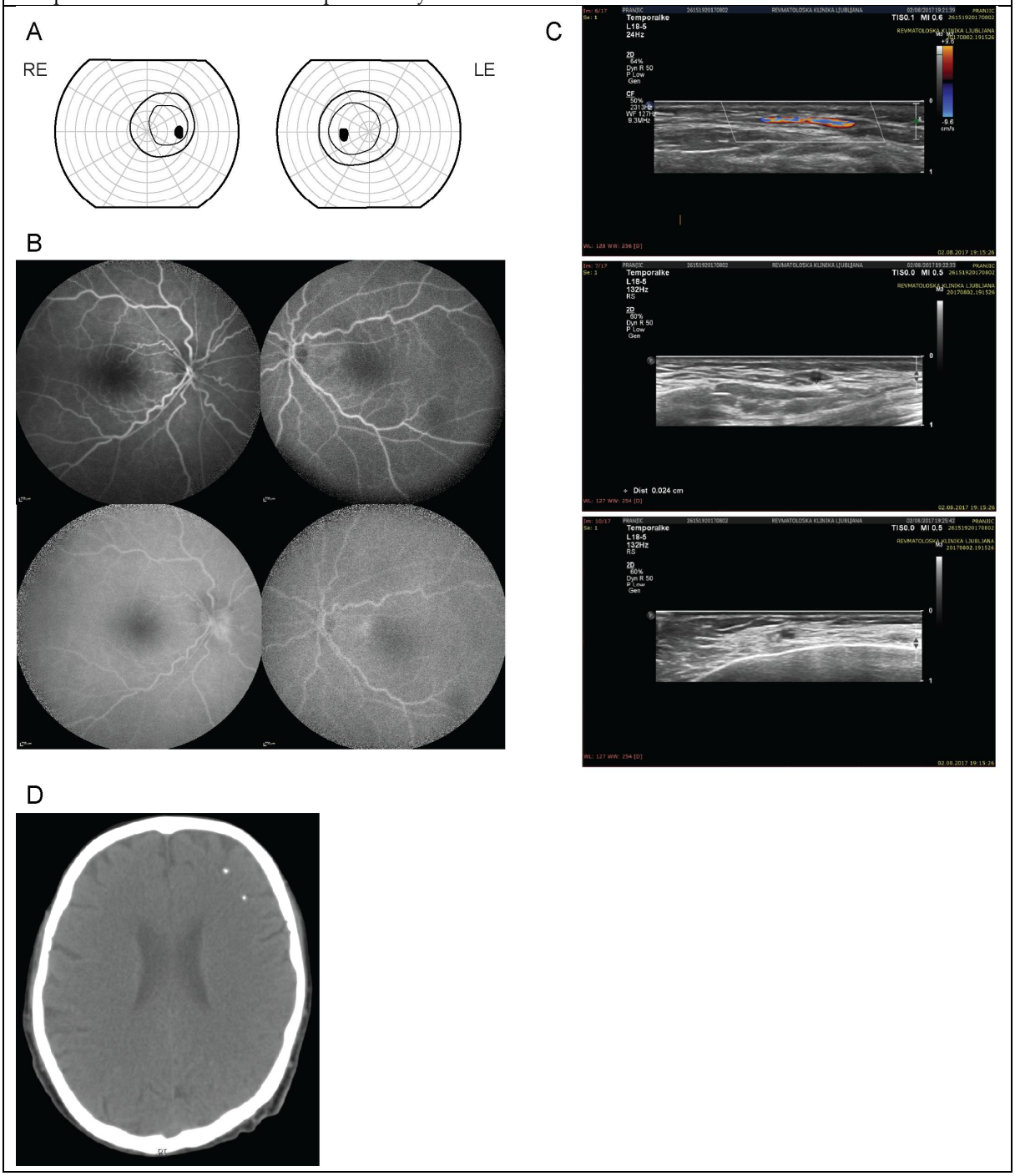
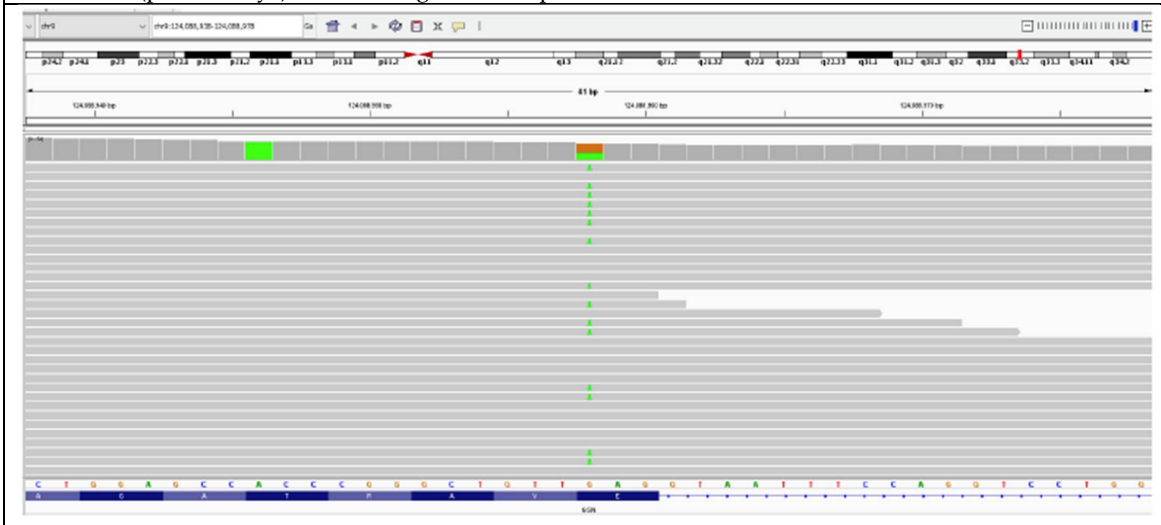
Supplementary Materials:

Supplementary Figure S1. A. Goldmann visual field of the patient II-1 showing concentric bilateral constriction of the visual fields, with non-detectable isopters V1 and V2. B. The fluorescein angiography of patient II-1. There were normal vessel filling times with leakage of contrast from the right optic disc in the late phase. C. The CT of the head of patient II-1 showing diffuse calcifications of the nucleus pallidum and nucleus caudatus. D. Ultrasound of the temporal arteries: On the main, frontal and parietal branch of temporal artery no halo sign was seen and the compression sign was negative. Thickness of intima-media complex on frontal branch of temporal branch of temporal artery was 0.024 cm.



Supplementary Figure S2. Loose skin (cutis laxa) in patients II-1 (A, F), II-2 (B), II-3 (C), II-8 (D) and II-5 (E). Note that all affected family members shown on the picture (II-1, II-2, II-3, II-8) exhibit lower lid dermatochalasis, whereas the family member II-5 carrying the wild type of GSN gene does not.

Supplementary Figure S3. Next generation sequencing revealing a novel heterozygous variant c.1738G>A (p.Glu580Lys) in the *GSN* gene in the proband.



Supplementary File 1. Questionnaire

Eye Clinic, University Medical Centre of Ljubljana
Grabovičeva 46
1000 Ljubljana
Slovenia

Dear Sir or Madam,

Genetic analysis of the DNA of your relative has revealed a novel variant of mutation in the gelsolin gene, which is associated with a rare disorder called gelsolin amyloidosis (Meretoja syndrome). This disorder may affect the eyes (cornea), skin, kidneys and heart, and it usually affects several relatives in the same family.

We kindly invite you to participate in further analysis of the novel mutation by sending us a saliva sample. The aim is to document all mutation carriers in your family and offer you counselling regarding possible further healthcare measures and the risk for mutation in your children. Genetic analysis is possible even if you do not exhibit any symptoms or signs of the disease.

In case you are willing to participate in the genetic analysis, please find enclosed a kit for genetic material. Please follow the enclosed instructions for material harvesting. Together with the signed consent form and filled out questionnaire, please send it back to the address written on the enclosed envelope. You will be informed about the genetic examination results within six to twelve months.
In diagnostics, clinical evaluation is fundamental and, when that is not available, images are a great substitute. If possible, please enclose an image of your face and/or eyes and any healthcare documentation.

Please find enclosed:
- Genetic material harvesting kit
- Questionnaire regarding impairment of different organ systems
- Consent form for genetic testing

Do not hesitate to contact us if you have any questions:
ana.fakin@kclj.si
majc.potrc@gmail.com

NAME AND SURNAME OF PARTICIPANT:
ADDRESS:
BIRTH DATE:
Relation to ***:

a. Sister
b. Brother
c. Son
d. Daughter
e. Other: _____

QUESTIONNAIRE

Please fill out the following 6 questions with YES or NO or OTHER.

1. Do you have any changes on your corneas? YES NO

   If yes, at what age did ocular changes appear? _____
   Do you have any other eye problems or disease? _____

2. Do you have any heart problems? YES NO
   If yes, what kind of problems do you have? _____
   At what age did the problems appear? _____

3. Do you have any kidney problems? YES NO
   If yes, what kind of problems do you have? _____
   At what age did the problems appear? _____

4. Have you had facial nerve impairment (loss of blinking control, decreased tearing, drooping of the mouth on the affected side, altered sense of taste)? YES NO
   At what age did the changes appear? _____

5. Have you noticed thickened facial or eyelid skin?
   At what age did the changes appear? _____

6. Do you have any other health problems? YES NO
   If yes, which problems do you have? _____