

- **Register för retinala degenerationer**

*Patienternas register 98% undersökta
SUS/Lund*

- **Brown-Vialetto-Van Laere syndrome-2**

*Progressiv dövblindhet ataxi vid brist på
riboflavin*

- **Vitamin A terapi vid retinitis pigmentosa**

*Samlad vetenskaplig fakta om denna terapi
samt informationsblad*

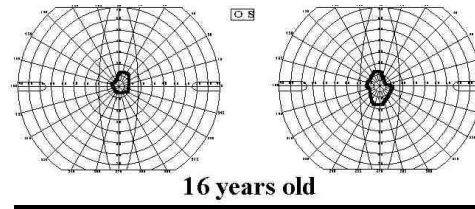
Sten Andréasson



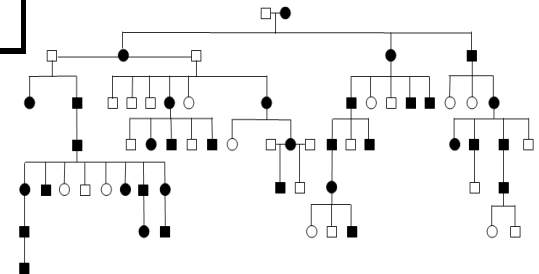
Register för ärftliga näthinnesjukdomar 1990 – 2018

3500 patients 98% undersökta spec mott ögonkliniken SUS

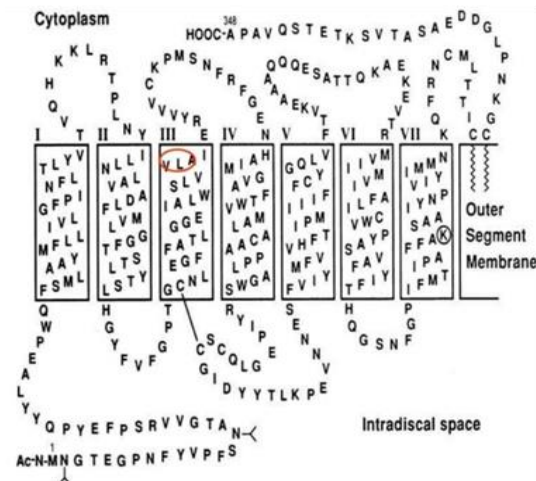
DNA bank > 3000 skriftligt godkännande av var patient



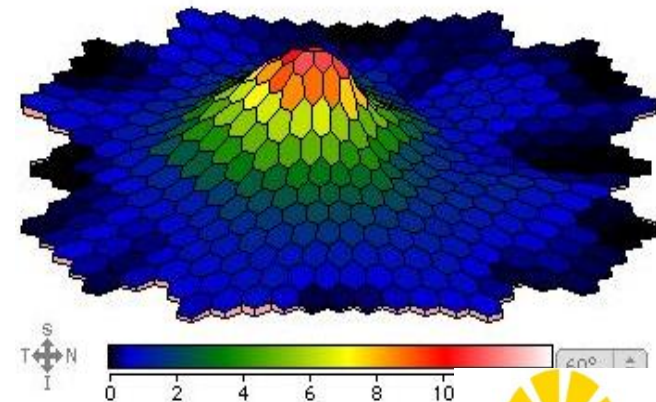
Pedigree



Genotype/DNA biobank



Phenotype/ Electrophysiology





Översikt av resultat från familjer från Svenska RP-registret

- **ARRP - autosomal rec RP**
- **ADRP - autosomal dom RP**
- **XLRP – könsbunden RP**
- **LCA – tidig form av RP**
- **CORD – omvänd RP**
- **ACHM – avsaknad av tappfunktion**
- **VMD2 o STGD1 – initialt påverkan av makula, gula fläcken**
- **XLRS – Skikt bildningen i näthinnan retinoschis**
- **USH – Synhörsel handikapp**
- **Övriga syndrom**

Översikt av resultat från familjer från Svenska RP-registret

- LCA – tidig form av RP

RP 27 - *NRL*

LCA 1 - *GUCY2D*

LCA 2 - *RPE65*

LCA 3 - *SPATA7*

LCA 4 - *AIPL1*

LCA 8 - *CRB1*

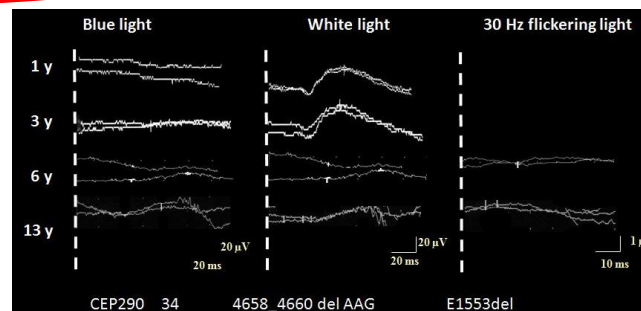
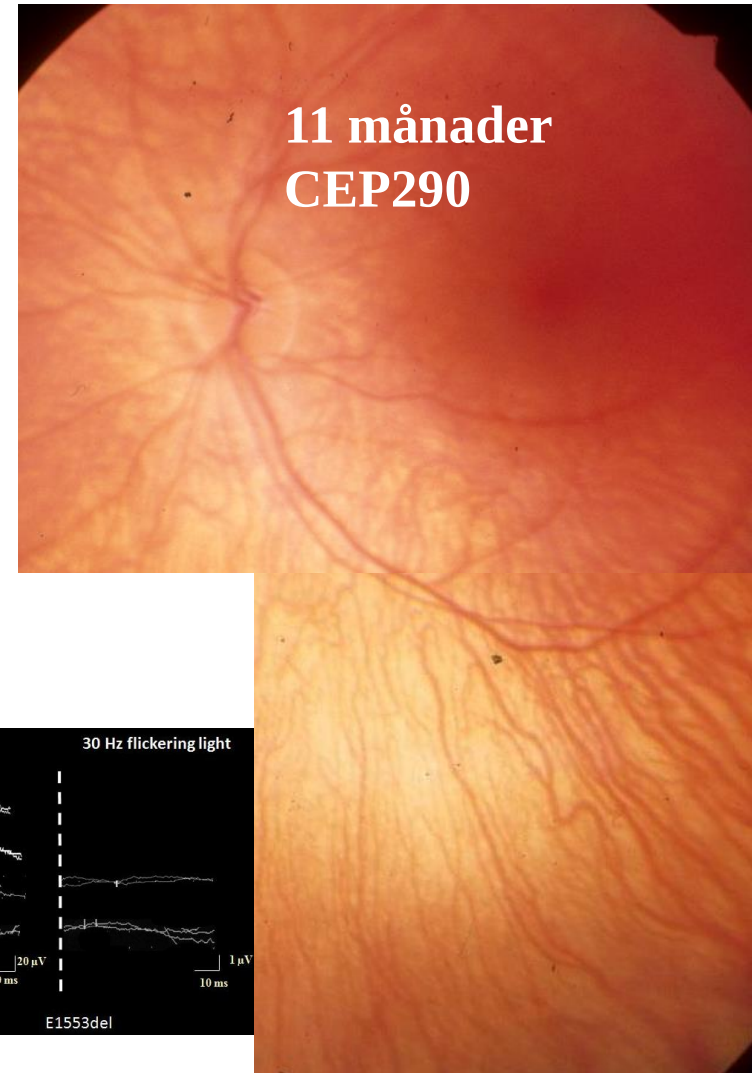
LCA 10 - *CEP290*

LCA 13 - *RDH12*

RP 1 – *RP1*

LCA 10 - *CEP290*

11 månader
CEP290

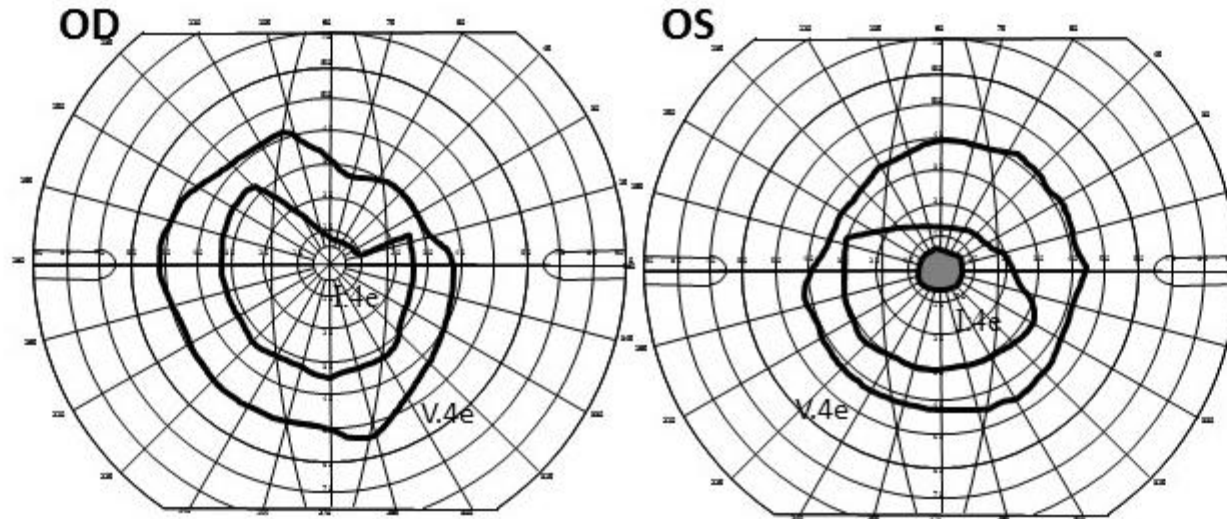


Uppföljning - Register retinala degenerationer – **Vitaminer B12** *forskning*

1992

9 yrs

Visual Field Goldman



**Best corrected
visual acuity**

0.15/FC2.5m

2016

Not measurable. Some residual field in periphery

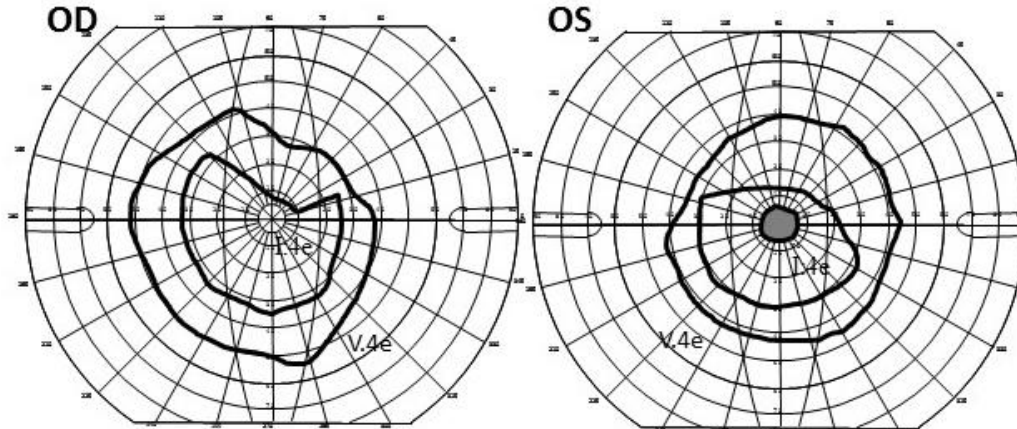
FC 0.5m/FC 0.5 m

- Brown-Vialetto-Van Laere syndrome-2 (BVVLS2)
- Progressiv ataxia,
- Synnedsättning
- Grav hörselnedsättning
- muskelsvaghet,
- Ibland andningsproblem
- *SLC52A2*, som kodar för riboflavin transporter 2 (RFVT2).

Visual Field Goldman

Best corrected
visual acuity

1992



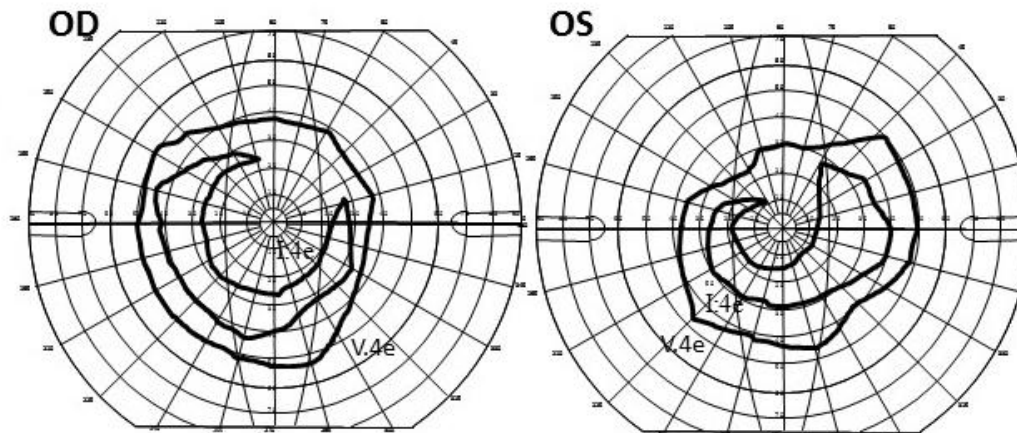
0.15/FC2.5m

2016

Not measurable. Some residual field in periphery

FC 0.5m/FC 0.5 m

2018



0.16/0.07



Oral therapy for riboflavin transporter deficiency - What is the regimen of choice?

Gorcenco S, Vaz FM, Tracewska-Siemiatkowska A, Tranebjærg L, Cremers FPM, Ygland E, Kicsi J, Rendtorff ND, Möller C, Kjellström U, Andréasson S, Puschmann . Parkinsonism Relat Disord. 2018 Oct 15. pii: S1353-8020

Vitamin A FFB 2018

A-vitaminbehandling vid RP

Svenska RP-föreningen

<https://retinanytt.se/a-vitaminbehandling-vid-rp/>

Vitadral droppar, 30,2 mg/ml (retinolpalmitat), förpackning 50 ml. Licenspreparat.

Godkänd av läkemedelsverket som licenspreparat

Enda behandling, som vetenskapligt möjligen kan hindra progressen av RP.

1. A randomized trial of vitamin A and vitamin E supplementation for retinitis pigmentosa. Berson EL, Rosner B, Sandberg MA, Hayes KC, Nicholson BW, Weigel-DiFranco C, Willett W. Arch Ophthalmol. 1993 Jun;111(6):761-72

2. Effect of vitamin A supplementation on rhodopsin mutants threonine-17 → methionine and proline-347 → serine in transgenic mice and in cell cultures. Li T, Sandberg MA, Pawlyk BS, Rosner B, Hayes KC, Dryja TP, Berson EL. Proc Natl Acad Sci U S A. 1998 Sep 29;95(20):11933-8

3. Retinitis pigmentosa. Hartong DT, Berson EL, Dryja TP. Lancet. 2006 Nov 18;368(9549):1795-809. Review

4. Association of Vitamin A Supplementation With Disease Course in Children With Retinitis Pigmentosa. Berson EL, Weigel-DiFranco C, Rosner B, Gaudio AR, Sandberg MA. JAMA Ophthalmol. 2018 May 1;136(5):490-495



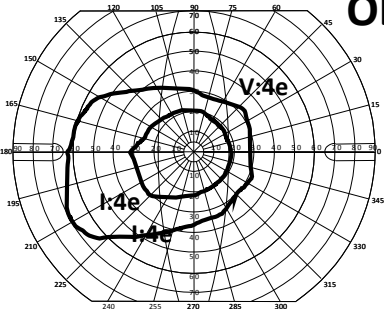
**UPDATE ON RESEARCH FOR RP TREATMENT:
VITAMIN A PALMITATE, OMEGA-3-RICH FISH
AND LUTEIN**

**FOUNDATION
FIGHTING
BLINDNES**

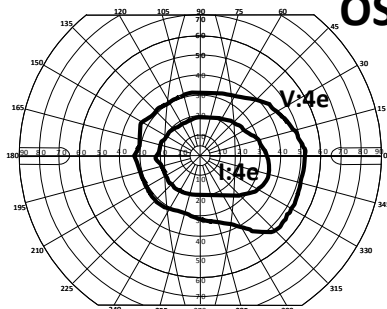


24 års uppföljning

OD

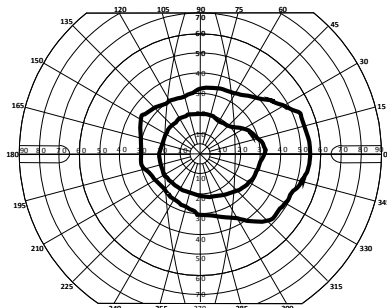
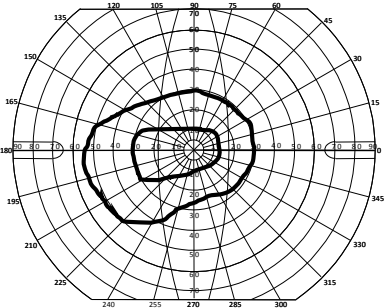


OS

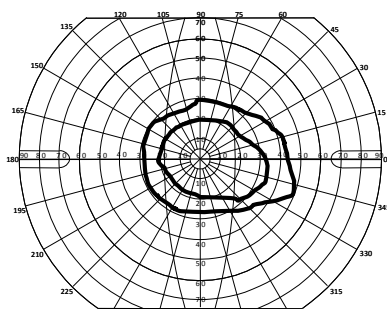
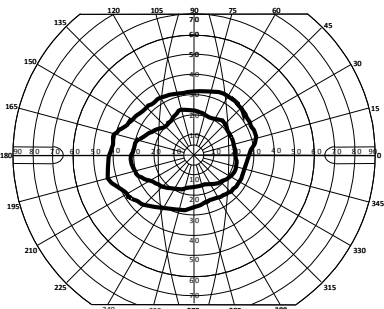


9 år

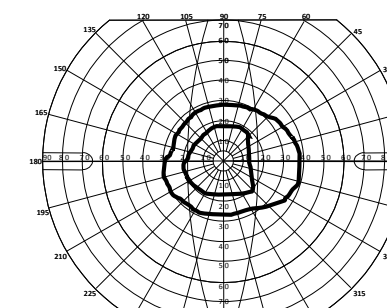
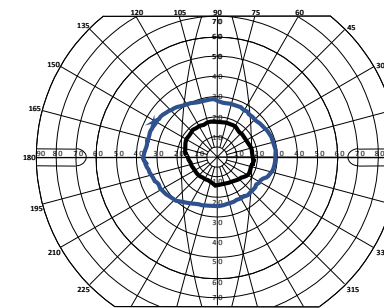
11 år



18 år



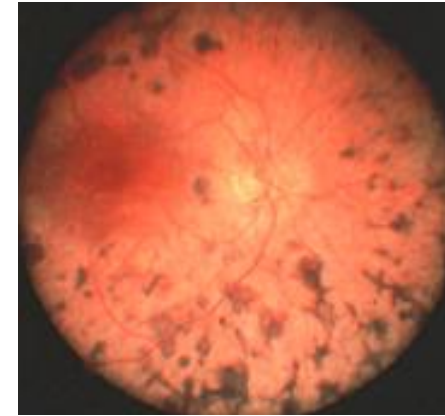
33 år



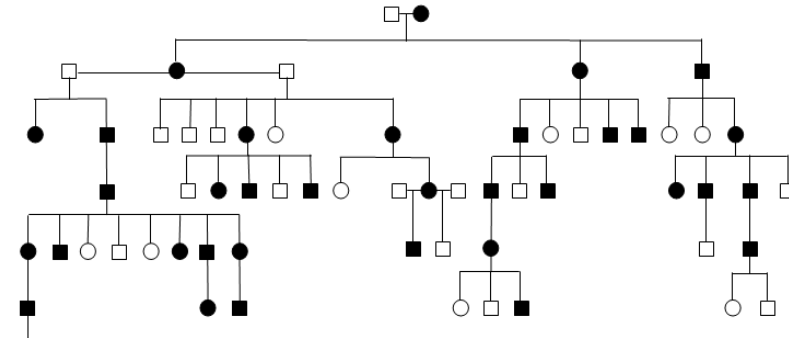
Rho Arg-135-Leu135 identifierad 1990



Son 15 år



mamma 45 år



A-vitaminbehandling vid RP Vilka skall behandlas?