Exudative Vitreoretinopathy (EVR)

aka. Familial Exudative Vitreoretinopathy (FEVR)
A disorder characterized by incomplete development of retinal vasculature. There is a wide variety of clinical appearances and severity but the pathology is the result of retinal ischemia. The disease is characterized by the development of hyper permeable blood vessels, neovascularization, vitreoretinal traction, retinal folds and retinal detachments.

Types

- Type 1 - Criswick-Schepens Syndrome
  - Autosomal Dominant
  - Gene: FZD4
  - Chromosome location: 11q14.2
- Type 2
  - X-linked recessive
  - Gene: NDP gene
- Type 3
  - Autosomal Dominant
  - Chromosome location: 11p13-p12 (approximately 30 cM from the EVR1 locus)
- Type 4
  - Autosomal Recessive
  - Gene: LRP5
  - Chromosome location: 11q13.2
- Type 5
  - Autosomal Dominant
  - Gene: TSPAN12
  - Chromosome location 7q31.31
- Type 6
  - Autosomal Dominant
  - Gene: ZNF408
  - Chromosome Location: 11p11.2
- Type 7
  - Autosomal Dominant
  - Gene: CTNNB1
  - Chromosome Location: 3p22.1

Resources

- Autosomal dominant exudative vitreoretinopathy. Ober et al. 1980
- Familial exudative vitreoretinopthy and related retinopathies. Gilmour DF. 2015

syndrome