Supplemental Data

Recessive mutations in *TSPAN12* cause retinal dysplasia and severe familial exudative vitreoretinopathy (FEVR)

Poulter et al. 2011.

Supplementary Figure 1. Additional clinical information for FEVR patient IV:1 in Family VL who is homozygous for the *TSPAN12* mutation p.L223P. A, This fundus photograph of the left eye taken at presentation (aged 3 months) shows a retinal fold traversing the macular region. B, Fundus photograph of the retinal fold shown in A focusing on the architecture of the retinal vasculature at the extreme temporal aspect of the fold. The retinal vessels appear to terminate with an abnormal configuration at the extreme temporal periphery and there is a small area consistent with intra-retinal haemorrhage inferior to the lower edge of the fold (Arrow). This patient also suffers from cystic fibrosis. Routine diagnostic screening of common UK *CFTR* mutations failed to identify any mutations in this patient. However, given that *CFTR* and *TSPAN12* are only 3 Mb apart on chromosome 7q31.1, it is highly likely that both mutations have arisen on the same ancestral chromosome and been co-inherited in this patient.
Supplementary Figure 2. Fluorescein angiography showing anterior avascularity of the left temporal retina in parents of the proband from family TM. A and B, father (Individual III:5). C and D, mother (Individual III:6). The posterior retina showed no abnormalities in either parent.