SUPPLEMENTAL FIGURES

The majority of autosomal recessive nanophthalmos and posterior microphthalmia can be attributed to biallelic sequence and structural variants in *MFRP* and *PRSS56*

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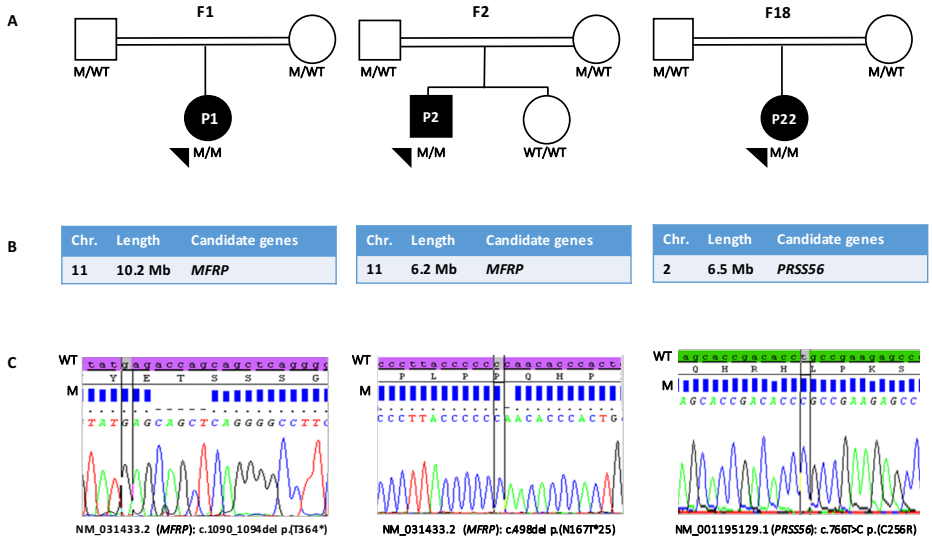
Supplementary Figure 1. Outcome of homozygosity mapping and targeted mutation screening in three consanguineous families



(A) Pedigrees of three consanguineous families (F1, F2 and F18). Filled symbols indicate affected individuals. Index cases are indicated by an arrow; a double line between a couple indicates consanguinity. Different genotypes are represented: homozygous mutant (M/M), heterozygous mutant (M/WT) and homozygous wild type (WT/WT). (B) Homozygosity mapping representing the size of the homozygous regions in which *MFRP* and *PRSS56* are located. (C) Sanger sequence electropherograms showing homozygous *MFRP/PRSS56* mutations.

Supplementary Figure 2. Molecular workflow in 21 unrelated families with NNO or MCOP.