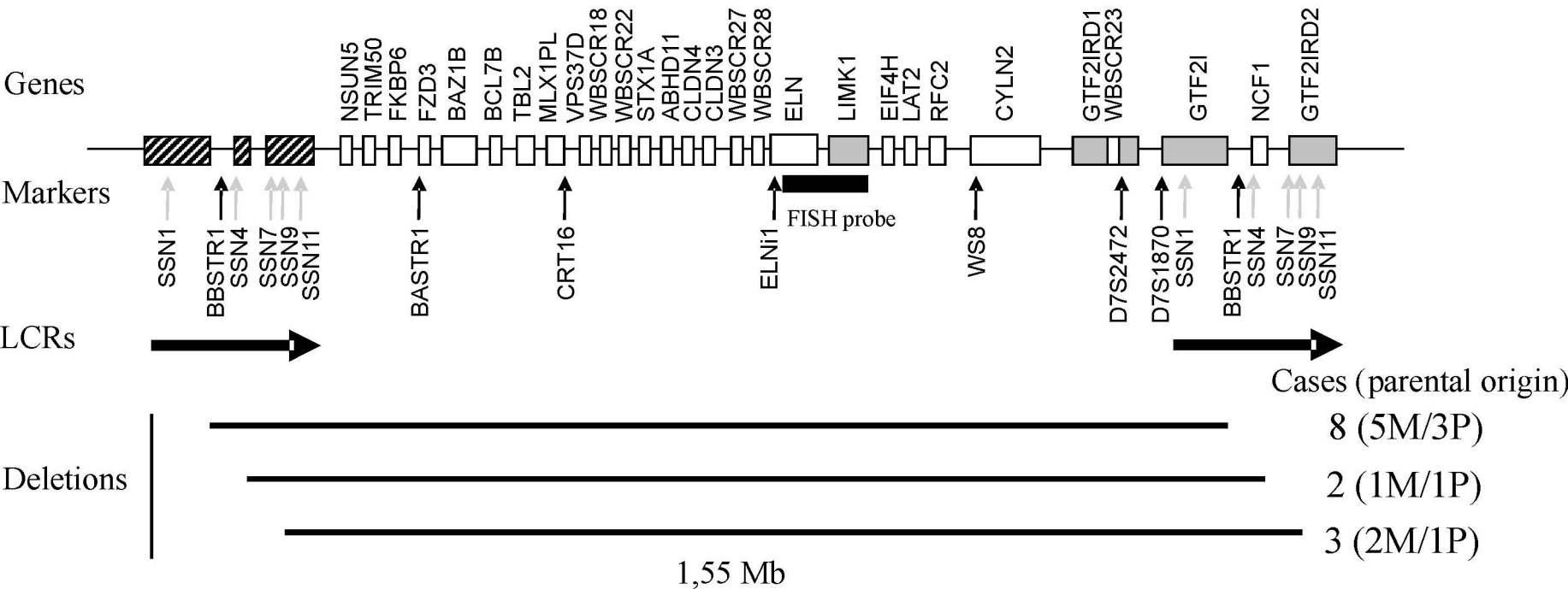


Supplementary Figure 1



Supplementary Figure 1

Schematic representation of the 7q11.23 region with its gene content. The location of the microsatellite markers (BBSTR1, BASTR1, CR16T, ELNi1, WBS8, D7S2472, D7S180) and paralogous sequence variants (SSNs1, 4, 7, 9 & 11) used in this study is indicated by black and grey arrows, respectively (20). The blocks of segmental duplications or low-copy repeats (LCRs) are represented by horizontal arrows. Hatched boxes on the centromeric LCR block represent pseudogene copies. Candidate genes for involvement in the visual phenotype based on previous data are shown as grey boxes. The extent of each type of deletion found in the patients is shown.