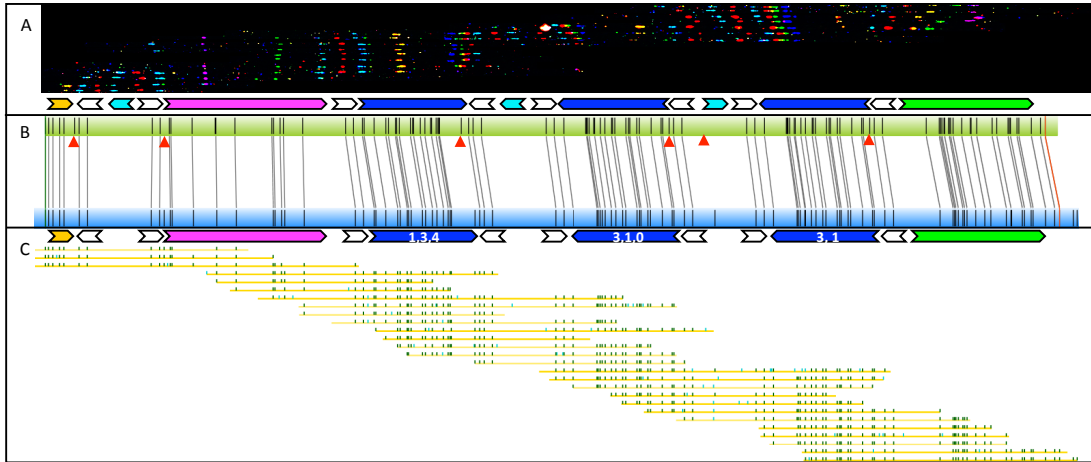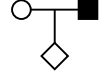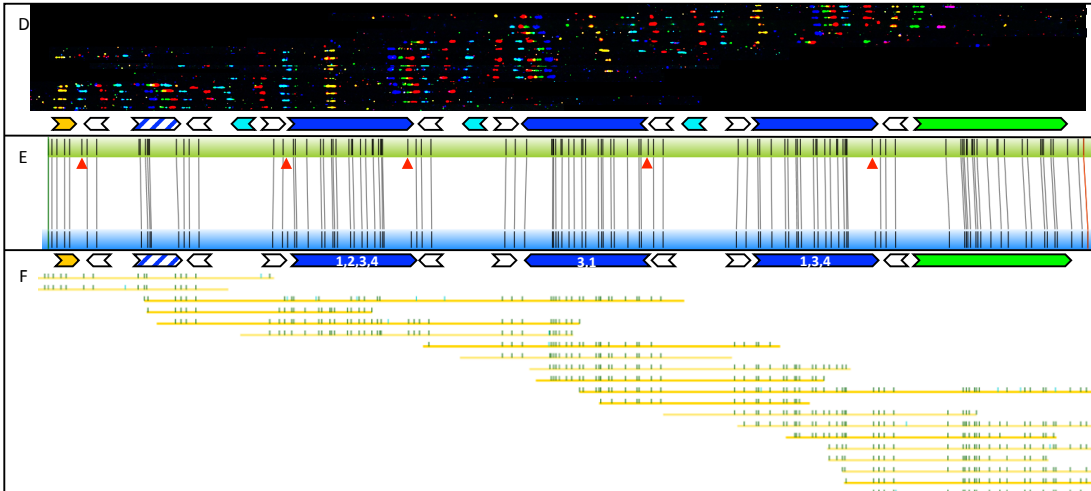KD77577567 – LCR22A Allele 1 - DLS
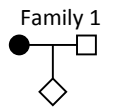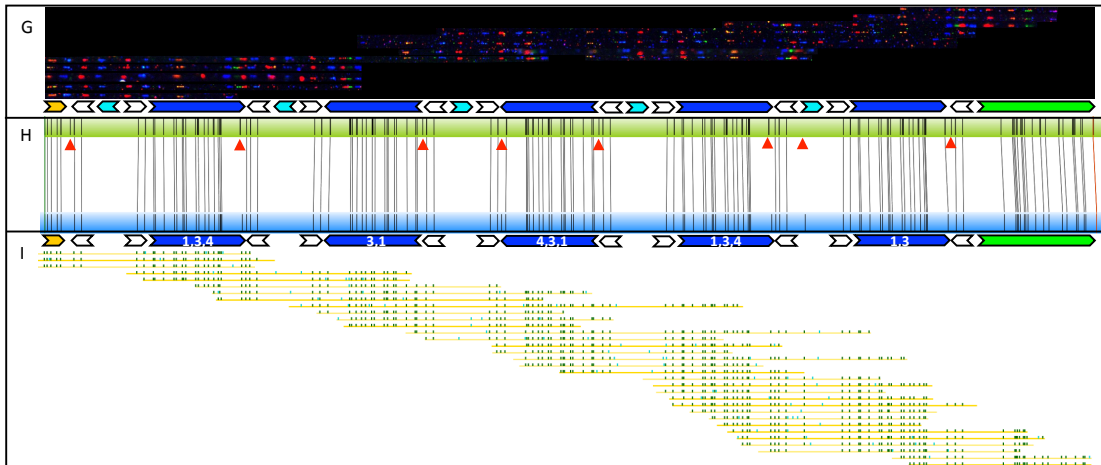

KD77577567 – LCR22A Allele 2 - DLS

Family 1
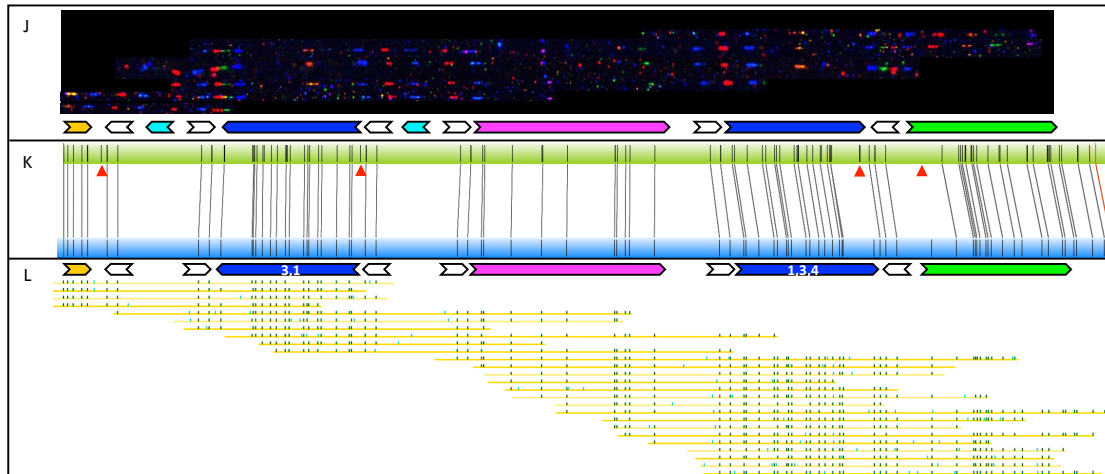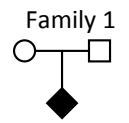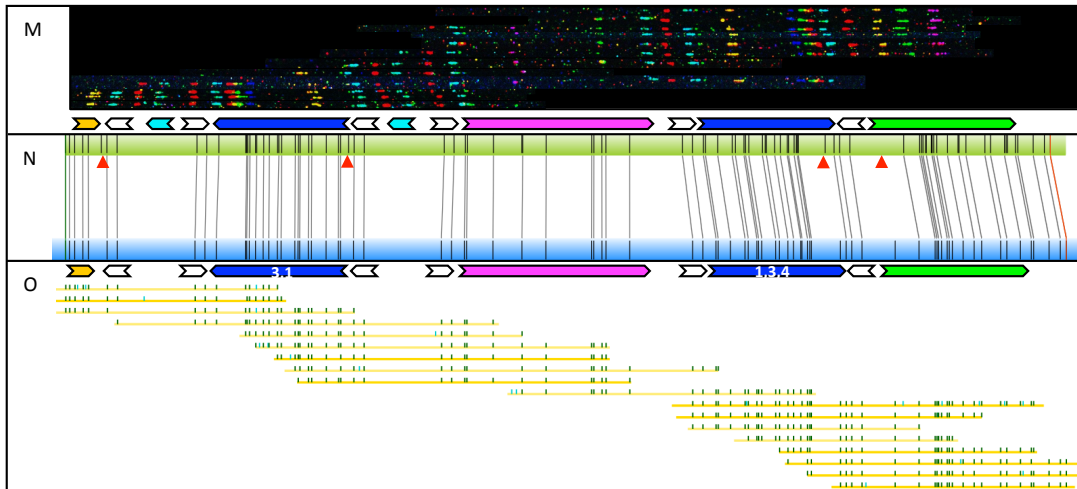
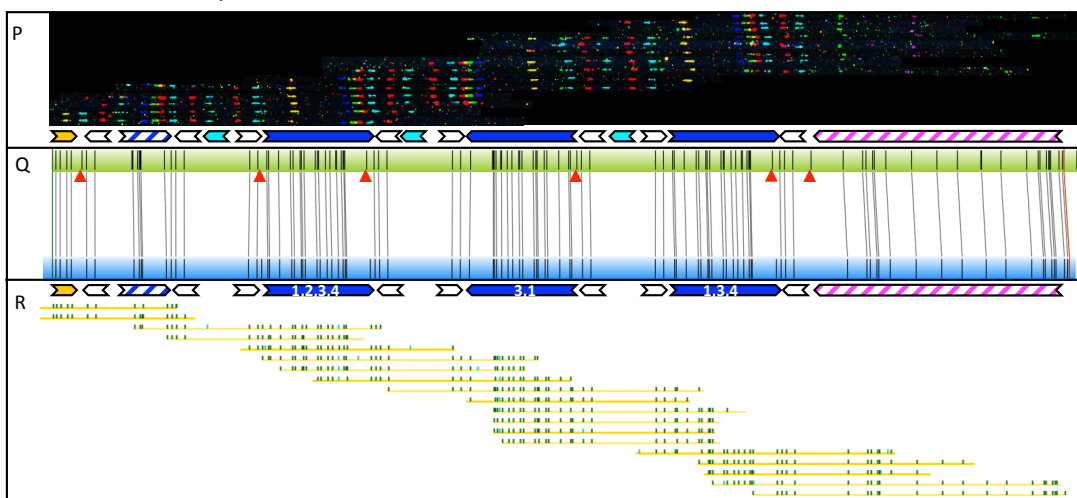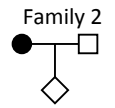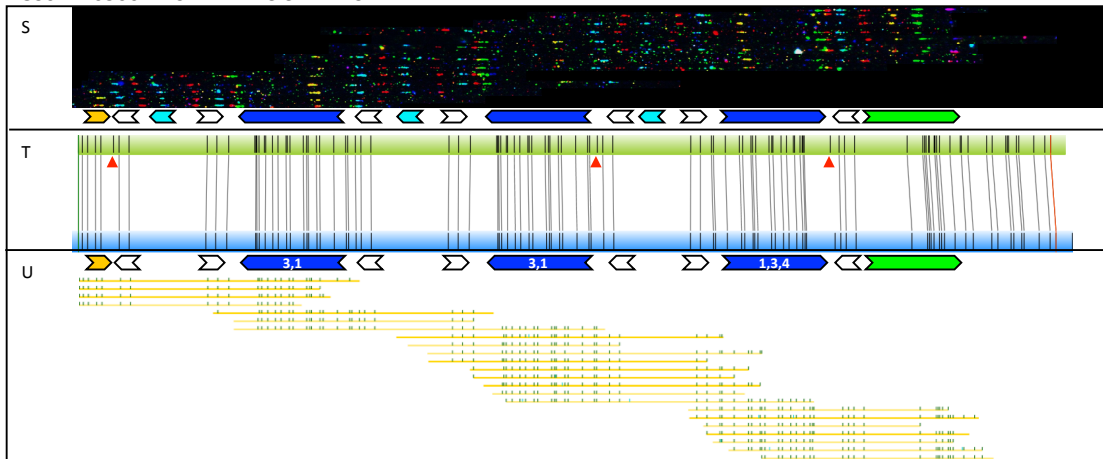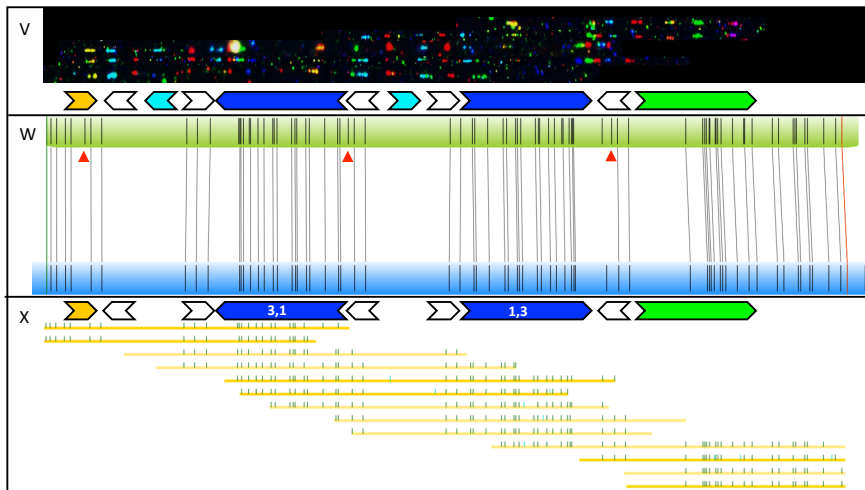KD70002217 – LCR22A Allele 1 - DLS

KD70002217 – LCR22A Allele 2 - DLS

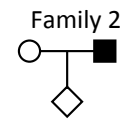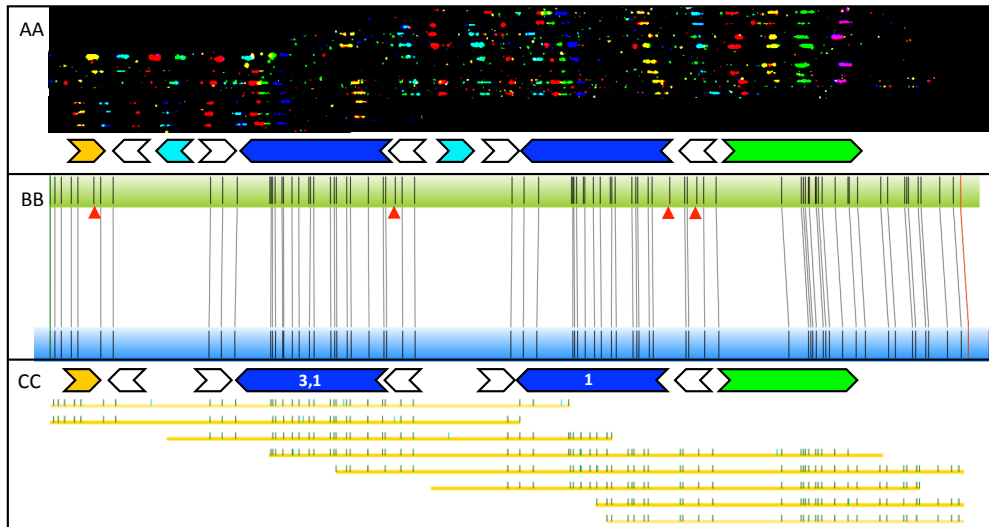

Family 1

SA85021 – LCR22A Allele 1

SA85021 – LCR22A/D

Family 1

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GS61116500 – LCR22A Allele 1 - DLS

Family 2

GS61116500 – LCR22A Allele 2 - DLS

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RA61116503 – LCR22A Allele 1 - DLS

Family 2

RA61116503 – LCR22A Allele 2 - DLS

RY263193 – LCR22A Allele 1 - DLS

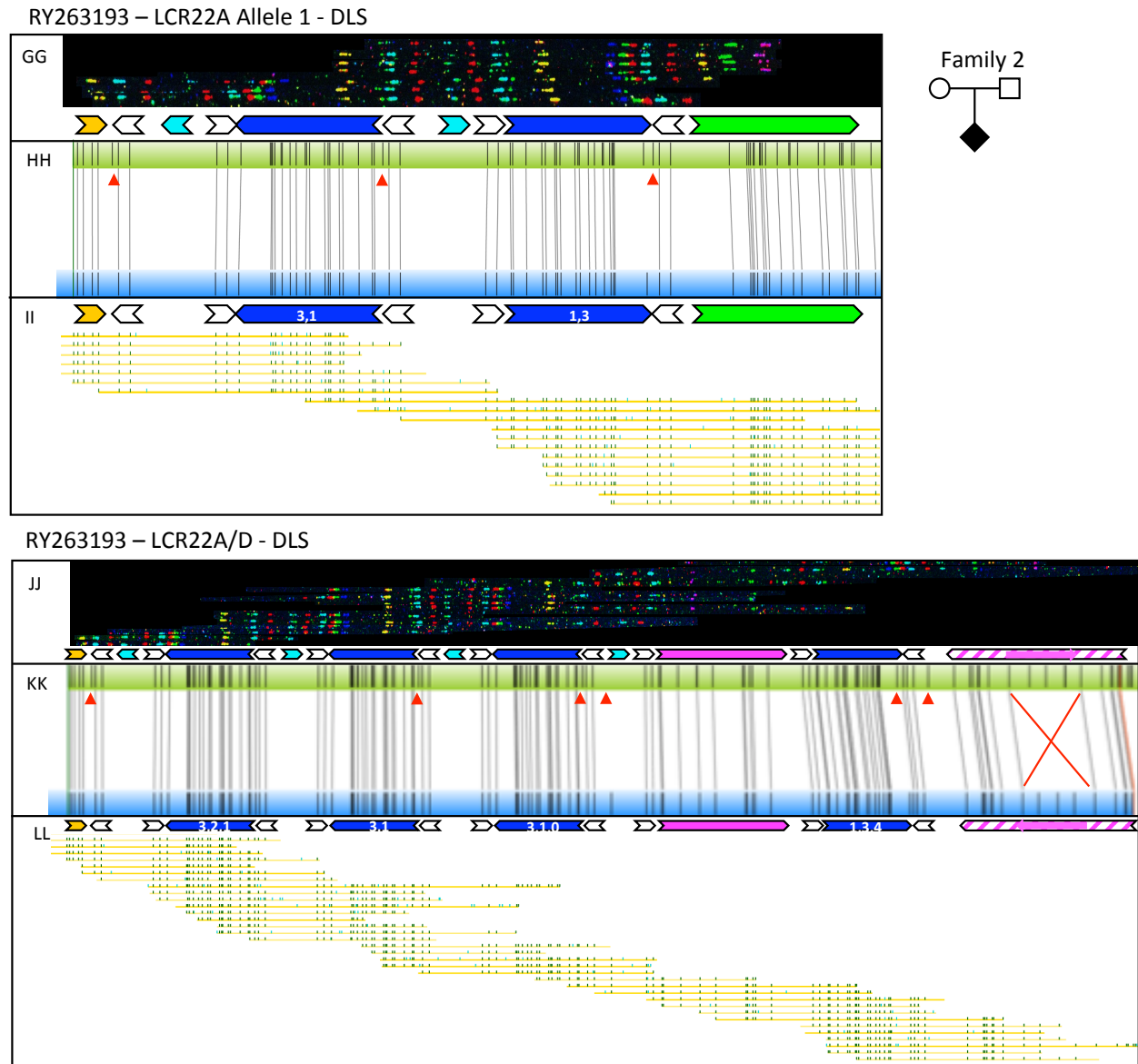**RY263193 – LCR22A/D - DLS**
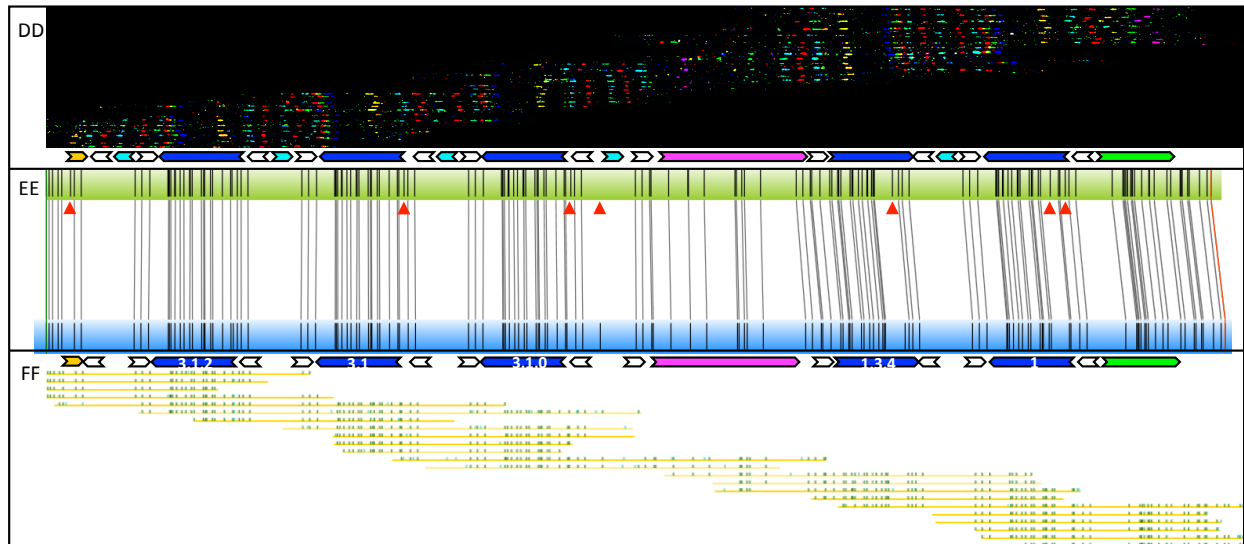
Family 2

Fig. S9.

Overlap of optical map and fiber FISH datasets in Family 1 and 2, two trios with one 22q11DS proband and both parents. Each family is indicated using a pedigree on the top right corner with the family member whose data is presented indicated by a filled circle or square. Both LCR22A alleles for each parent are shown. In the probands, the non-rearranged LCR22A allele and the rearranged LCR22A/D allele are shown. (A,D,G,J,M,P,S,V,AA,DD,GG,JJ) Top: Fiber FISH assembly, Bottom: its decomposition to duplicons as defined in Fig. 1.

(B,E,H,K,N,Q,T,W,BB,EE,HH,KK) Top: predicted optical map generated by *in silico* DLS labeling of hg38 duplicon sequences in the order observed by fiber FISH shown as green bar with black lines (DLS labels); Bottom: observed DLS optical map of the same allele shown as blue bar with black lines (DLS labels). Red arrowheads depict signal discrepancies between predicted and observed maps. (C,F,I,L,O,R,U,X,CC,FF,II,LL) Bottom: Optical map single molecules aligned to the observed LCR22A contig shown as yellow lines with green ticks (DLS

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labels); Top: decomposed to duplicons defined as in fig. 1 (top). Observed polymorphic DLS labels as defined in Fig. S6 are indicated by numbers 1-4 inside the duplicons.