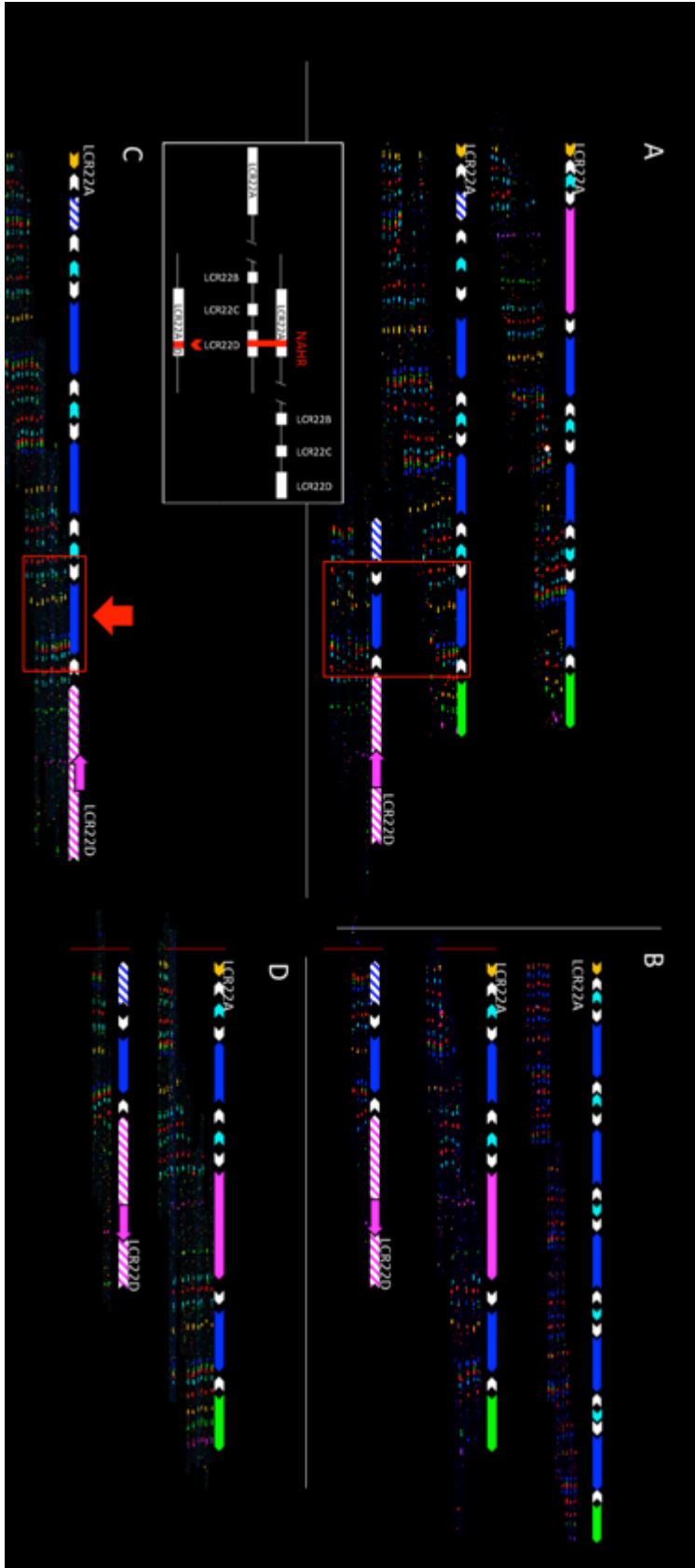
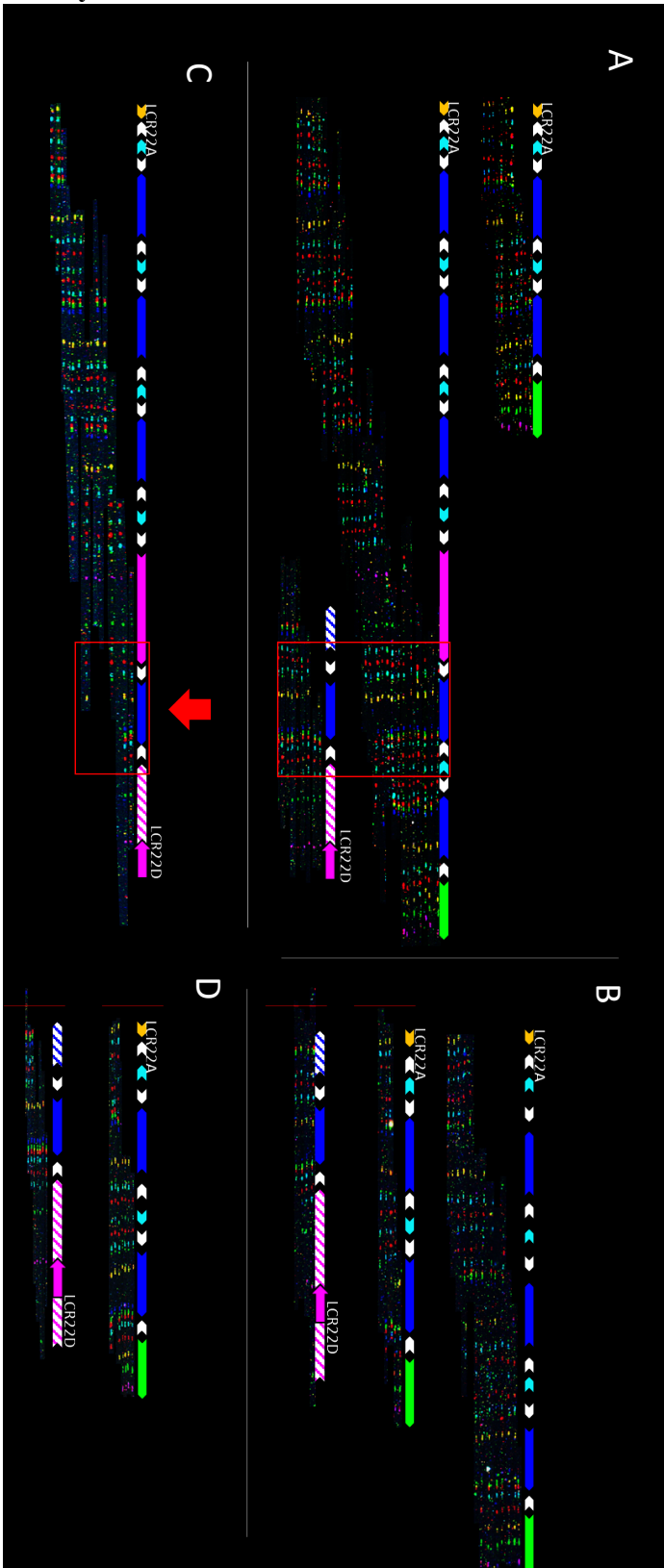


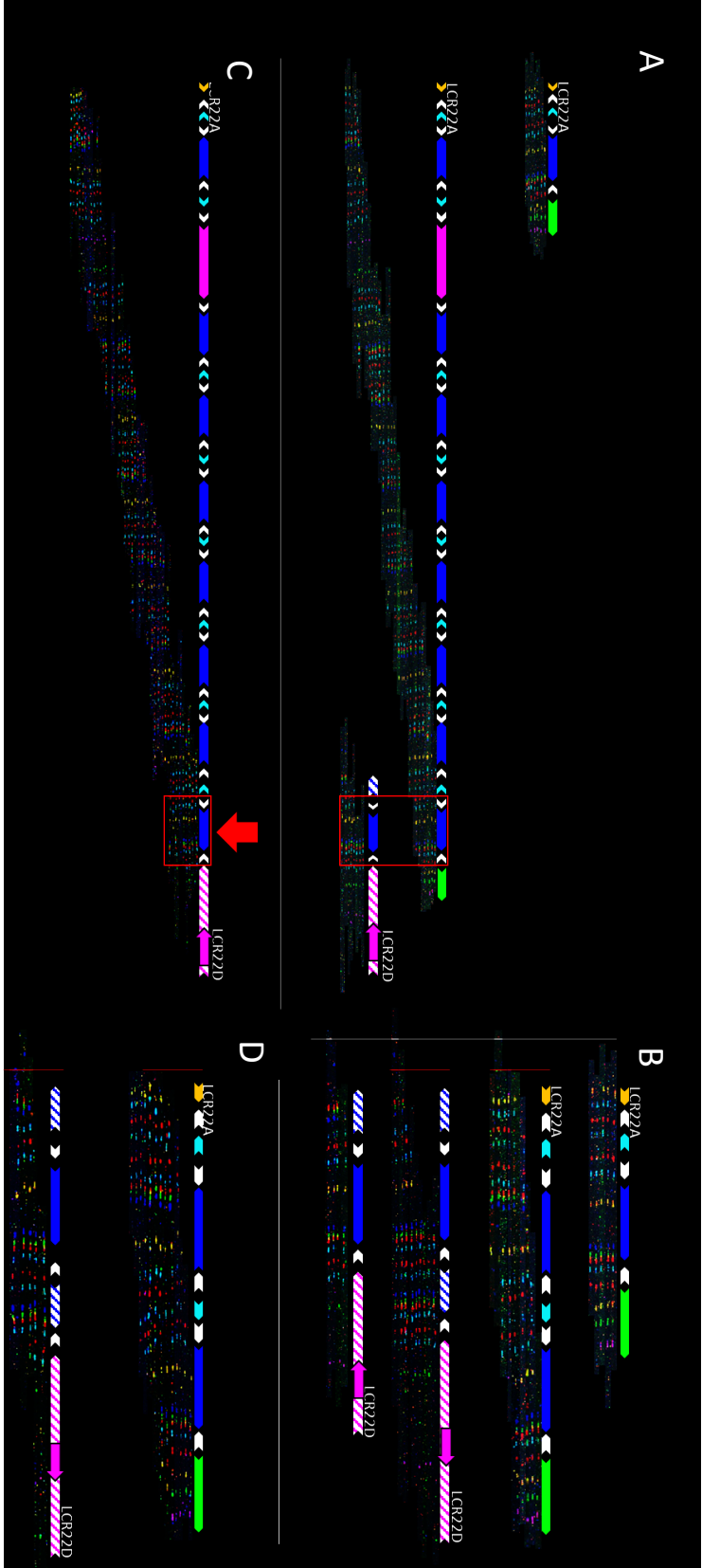
Family 1



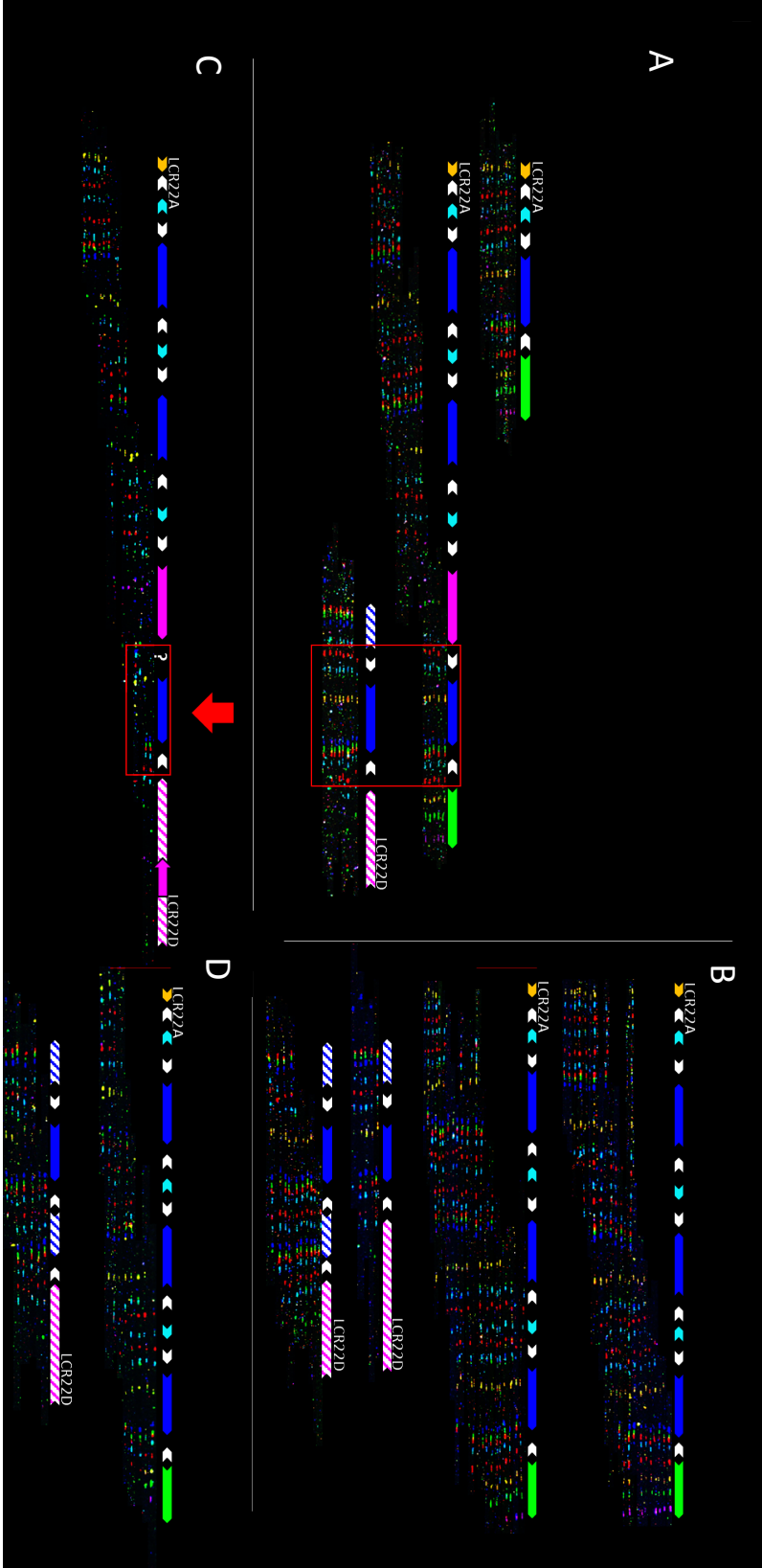
Family 2



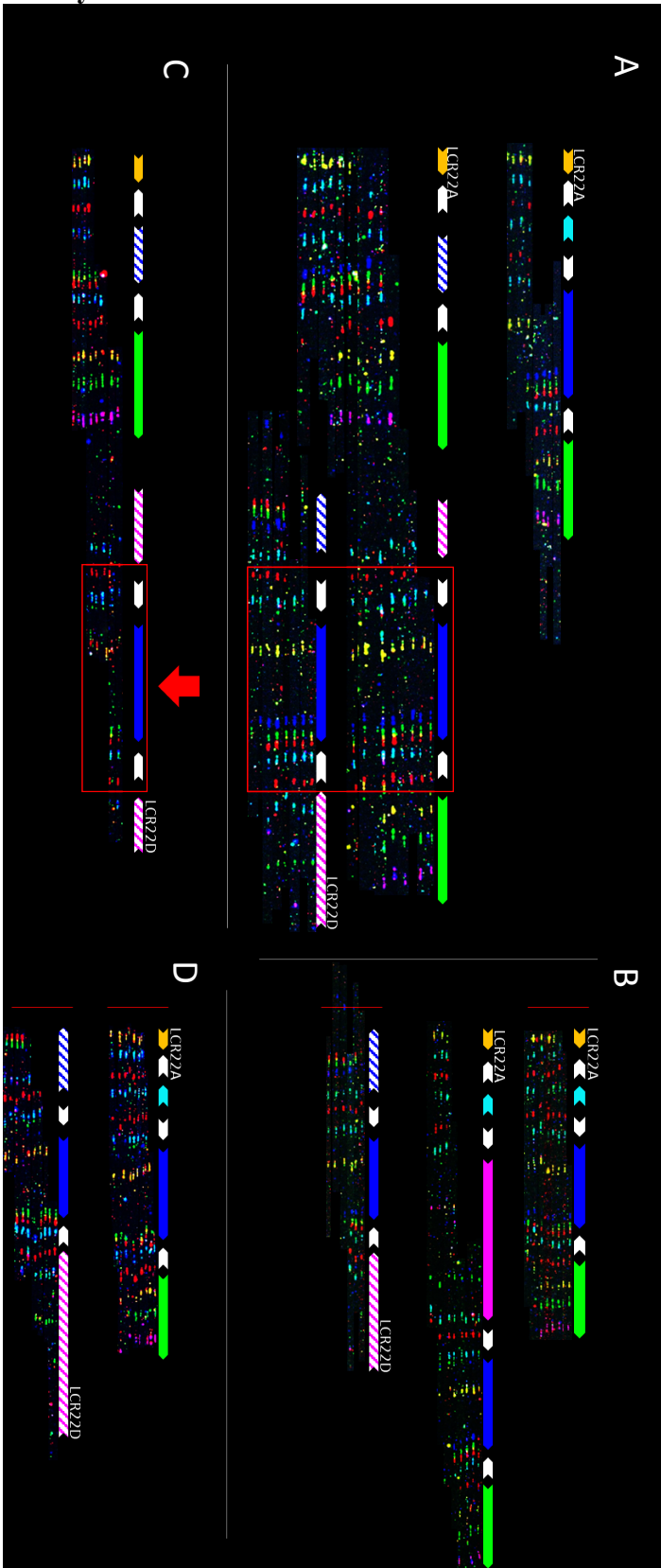
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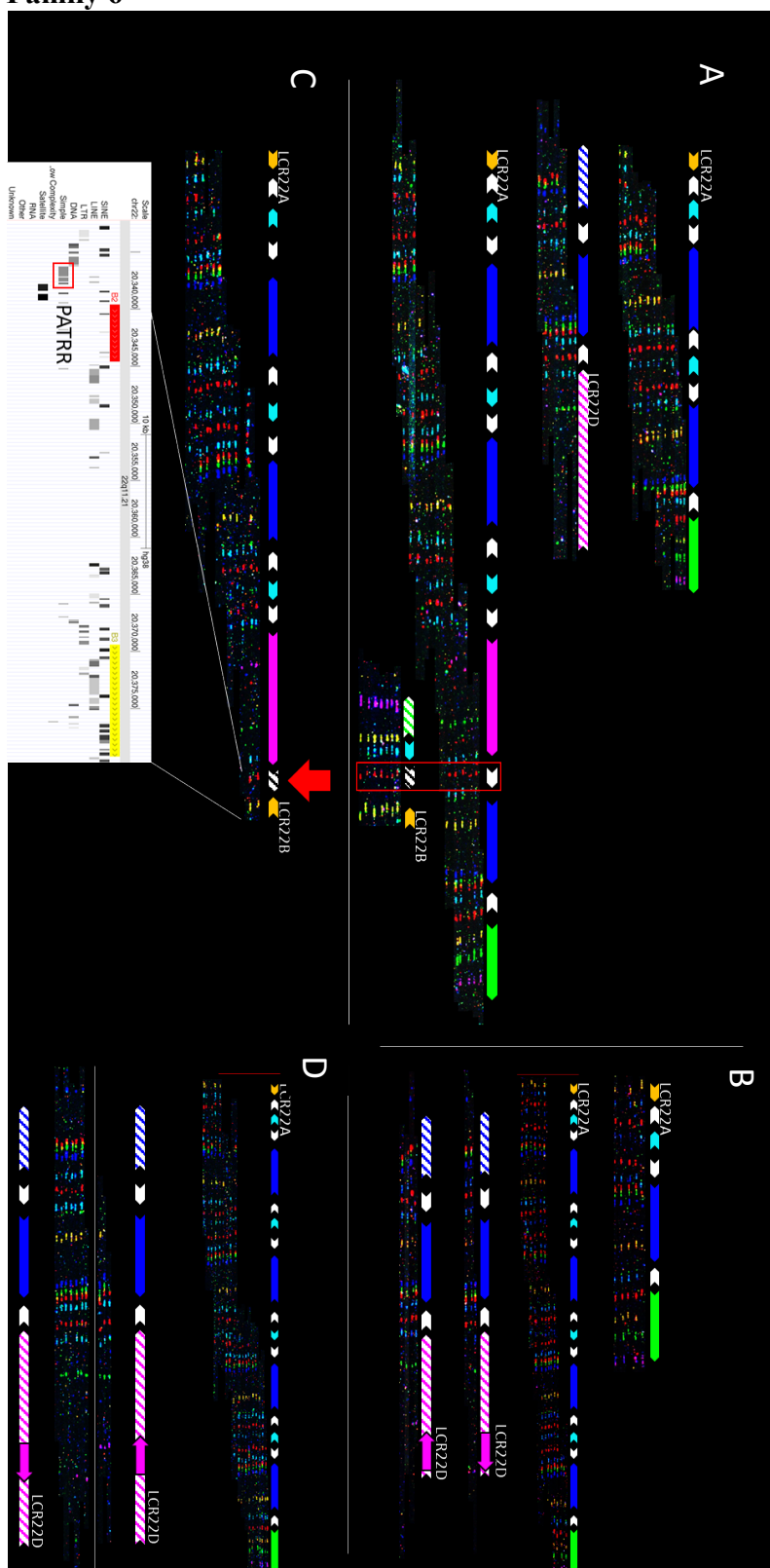
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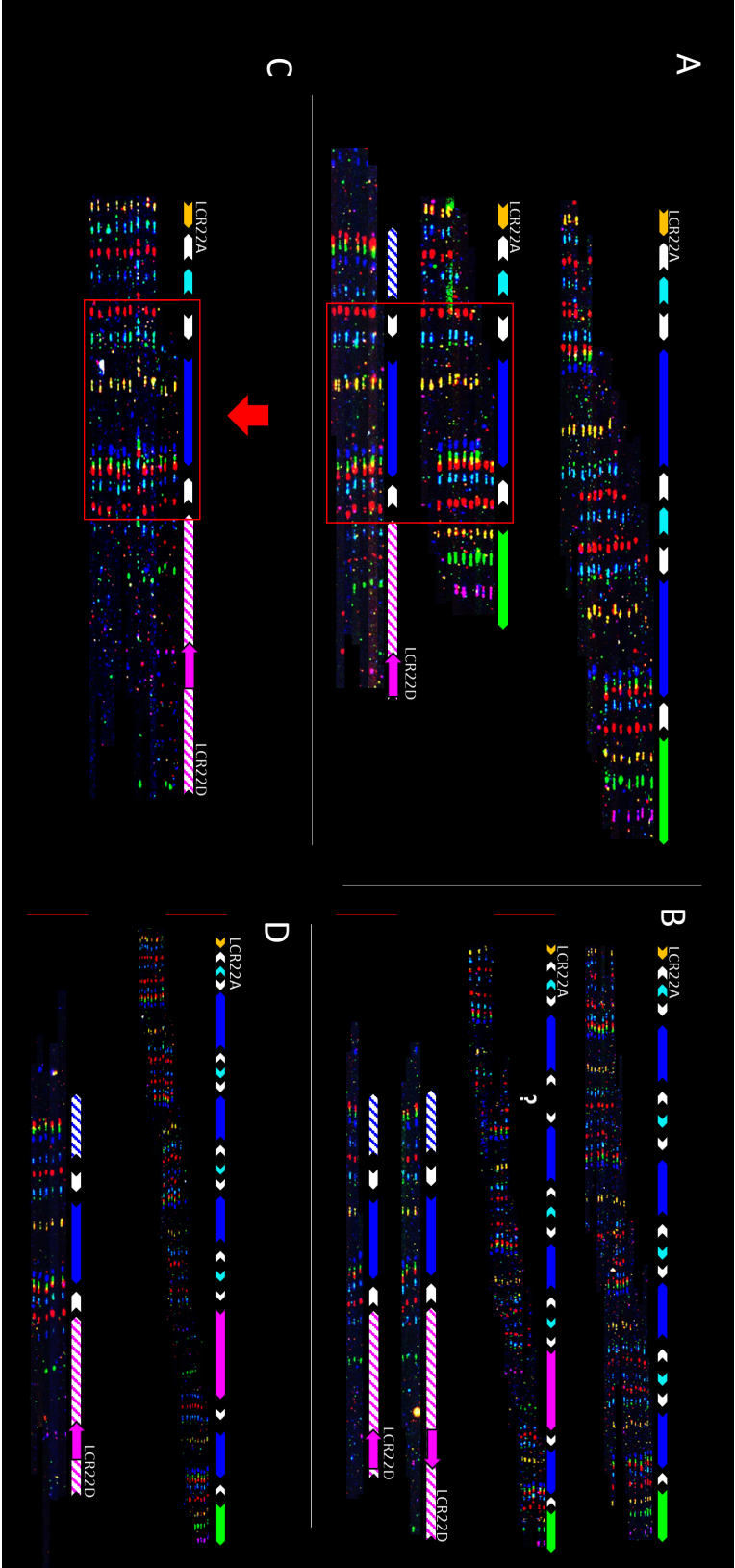
Family 5



Family 6



Family 7



Family 8

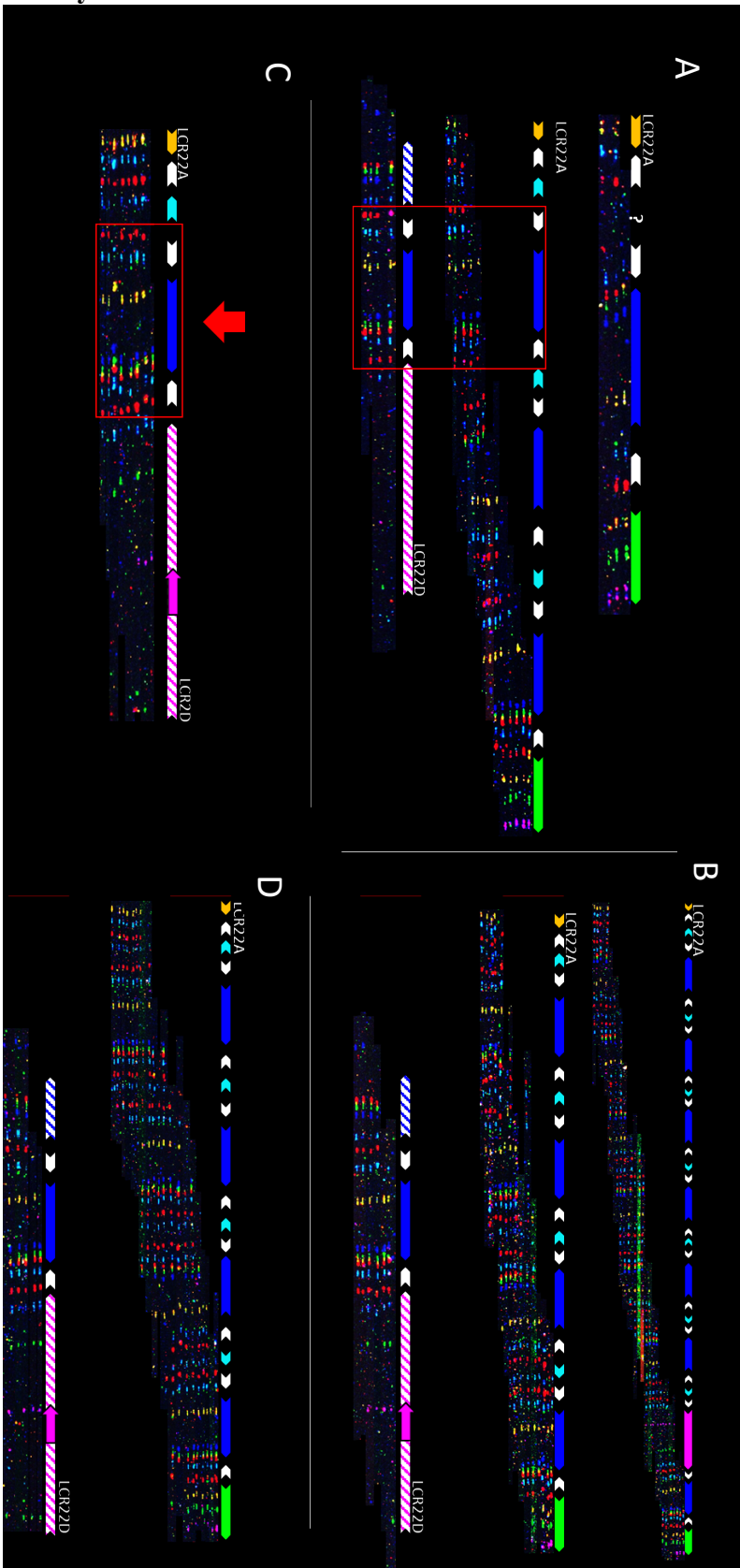


Fig. S7.

Analysis of the 22q11.21 deletion in a proband and parents using fiber FISH in eight families. Both alleles of LCR22A and LCR22D (or -B) are shown for each individual in the family trio. A) Parent of origin, B) other parent, C) proband rearranged allele, and D) proband non-rearranged allele. Fiber FISH assemblies are aligned with a duplicon representation as defined in Figure 1. In the parent of origin, the shared region between LCR22A and -D (or -B) is marked by a red box. Below misaligning alleles, the resulting rearranged LCR22 in the proband is displayed. A red arrow marks duplicon(s) that were involved in recombination and define the breakpoint region. The transmission of the un-rearranged allele is depicted on the right. LCR22A and -D alleles of the parent are displayed in the upper-right panel. Transmitted, non-rearranged parental alleles are marked by a red line. The lower right panel shows the non-rearranged alleles observed in the patient.