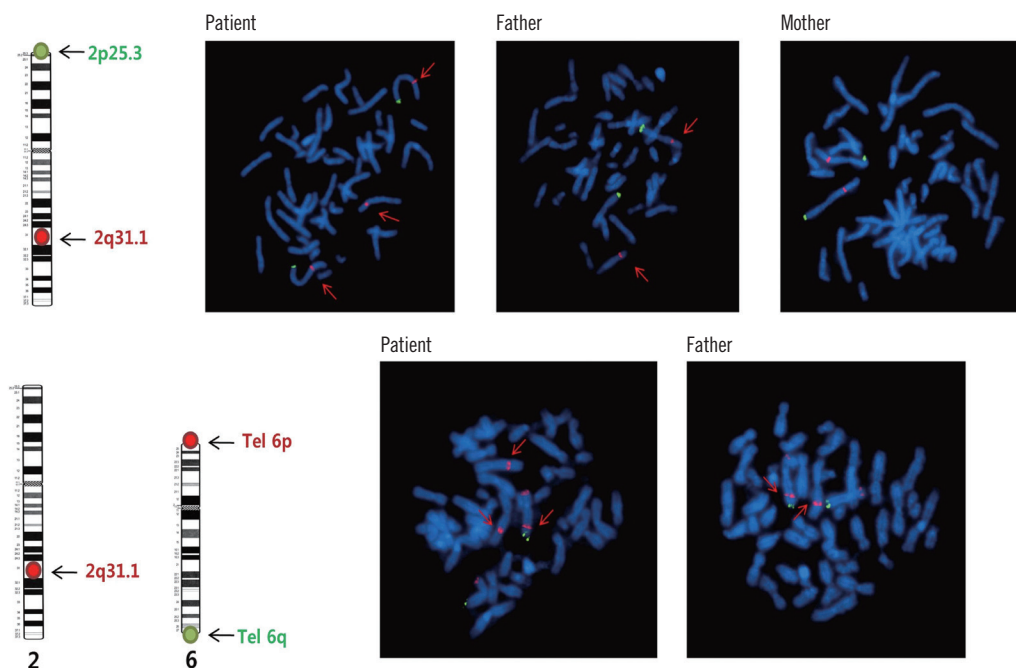
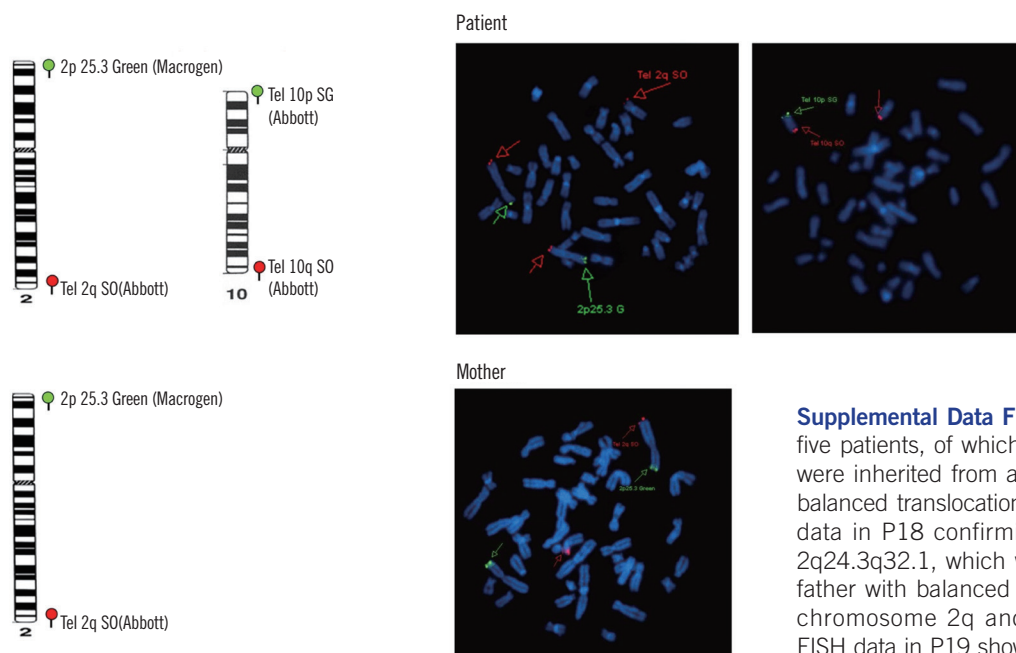


**A** 2q24.3-q32.1(163875703-183964083) 20Mb duplication  
ish der(6)ins(6;2)(CHR2:172375675-172855785)



**B** 2q36.3-q37.3(228622461-243028452) 14Mb duplication, 10p15.3(136361-2975072) 2.83Mb deletion  
ish der(10)t(2;10)(q36;p15)mat(10p006-,D2S447+)

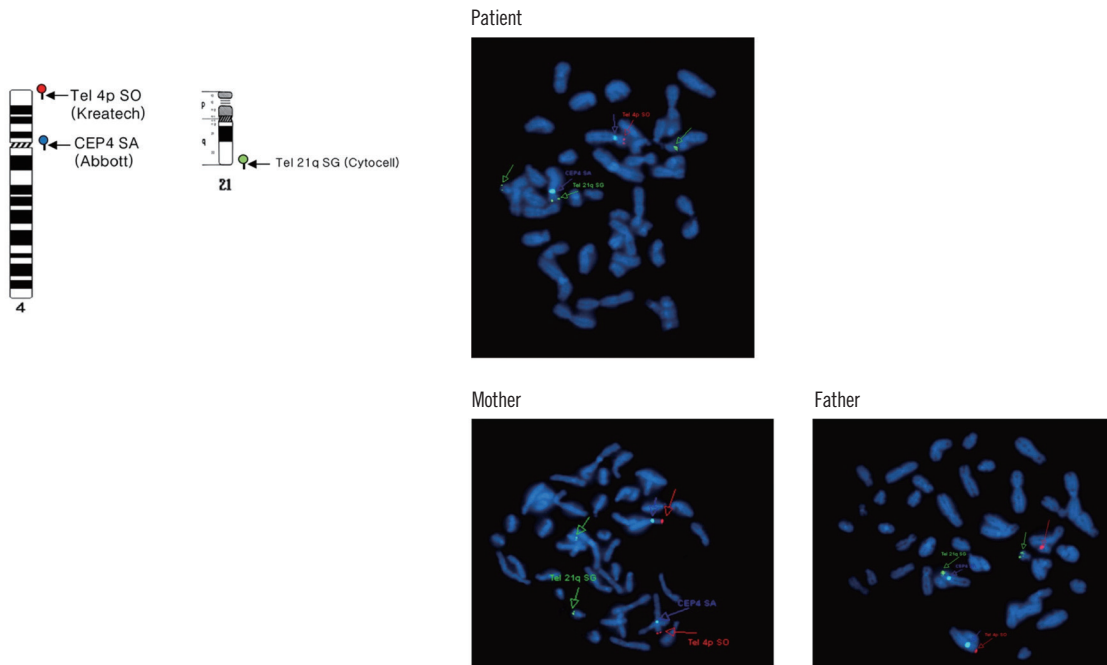


**Supplemental Data Fig. S1.** FISH data from five patients, of which the pathogenic CNVs were inherited from a healthy parent with a balanced translocation. (A) Metaphase FISH data in P18 confirming the duplication of 2q24.3q32.1, which was inherited from his father with balanced translocation between chromosome 2q and 6q. (B) Metaphase FISH data in P19 showing the 10p15.3 deletion and 2q36.3q37.3 duplication, which

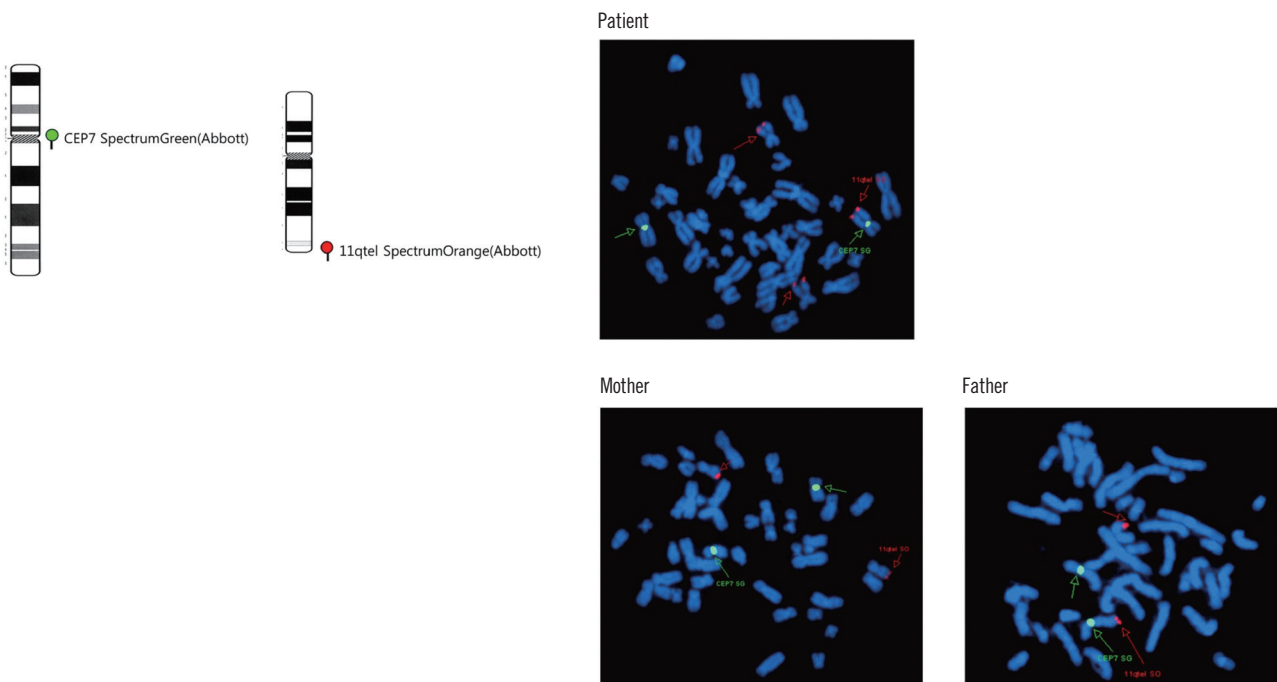
were inherited from his mother with balanced translocation between 10p and 2q. His father's FISH data are not shown here. (C) Metaphase FISH data in P30 confirming the 4p16.3p16.1 deletion and 21q22.2q22.3 duplication, which were inherited from his father with balanced translocation between 4p and 21q. (D) Metaphase FISH data in P46 confirming the 7q36.2q36.3 deletion and 11q24.2q25 duplication, which were inherited from his father with balanced translocation between 7q and 11q. (E) Metaphase FISH data in P95 showing the duplication of 19p13.3p13.2, which was inherited from his father with balanced translocation between chromosome 9q and 19p. His mother's FISH data are not shown here. Parental karyotyping was performed for the remaining three patients (P6, P37, and P89).

(Continued to the next page)

- C** 4p16.3-p16.1(71552-6164267) 6Mb deletion, 21q22.2-q22.3(41961820-48090317) 6Mb duplication  
ish der(4)t(4;21)(p16.1;q22.2)pat(4p022-,D4Z1+,63H24+)



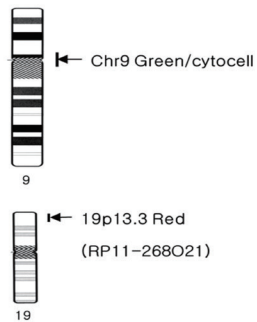
- D** 7q36.2-q36.3(153021783-159118707) 6Mb deletion, 11q24.2-q25(125128710-134927255) 9.79Mb duplication  
ish der(7)t(7;11)(q36.2;q24.2)pat(D7Z1+,D11S1037+)



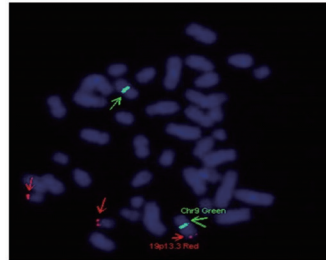
Supplemental Data Fig. S1. Continued.

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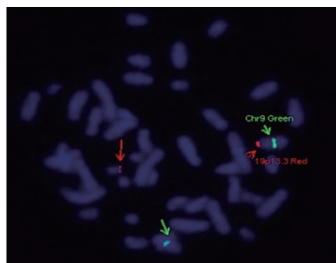
**E** 19p13.3-p13.2(266117-7599470) 7.3Mb duplication  
ish der(9)t(9;19)(q34.3;p13.2)(D9Z3+,RP11-268021+)pat



Patient



Father



Supplemental Data Fig. S1. Continued.