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Familial Robertsonian Translocation 13;21 in a Down Syndrome Patient with XYY/XY Mosaicism

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ABSTRACT Double aneuploidy involving XYY and trisomy 21 is rare. XYY/XY mosaicism has been described in only a single Down syndrome patient. The Robertsonian translocation t(13;21) is also rare among these individuals. We report for the first time the occurrence of t(13;21) in a mosaic XYY Down male. Analysis of GTG-banded metaphases revealed the karyotype of the propositus to be mos 47,XYY,der(13;21)(q10;q10),+21/46,XY,der(13;21),+21. Both his father and paternal grandfather were found to be carriers for the translocation. This 10-month-old child who presented with typical features of Down syndrome, developed leukemia and died at the age of 2¼ years.