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Prenatal Diagnosis of De Novo Reciprocal Translocation t(1;12)(q21.3;p11.2) with Trisomy 21 and Sperm FISH Analysis for Increased Aneuploidy Risk

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ABSTRACT Complex rearrangements such as de novo translocations together with aneuploidy are unusual situations in prenatal diagnosis. We report a case with de novo translocation t(1;12)(q21.3;p11.2) and trisomy 21. Father's sperm was analyzed for potential of increased risk of aneuploidy. Results showed no paternal increased risk for chromosomes 13, 18, 21, X, Y. Based on our results, we suggest that possible increased maternal aneuploidy risk and other possible mechanisms should be investigated to better understand cell division errors and to give better genetic counseling.