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复杂染色体重排的女性携带者与她健康的女儿 A de novo Complex Chromosomal Rearrangement Including Translocation on 1,5 and 12 in a Female Carrier with her Healthy Girl

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摘要

一例新生复杂染色体重排的女性携带者（complex chromosome rearrangement ,CCR），易位涉及1号、5号和12号染色体。病人因2次自然流产而要求进行外周血淋巴细胞G显带核型分析。最初G显带核型疑为46,XX,t(1;5;12)(1pter→1q25::12q24→12qter;5qter→5p11::1q25→1qter;12pter→12q24::5p11→5pter).经荧光原位杂交（FISH）技术检测，证实患者的核型为46,XX,t(1;5;12)(1pter→1q23::12q22→12qter;5qter→5p11::1q25→1qter;12pter→12q22::1q23→1q25::5p11→5pter).7年后病人再次妊娠，并拒绝产前诊断。女婴足月分娩，生长发育正常。核型为46，XX。比较以前报告的女性复杂易位携带者与我们报告的病例可以认为，CCR并不总是表现为自然流产或分娩畸形儿，仍有机会生出正常的孩子。Abstract: We reported in the paper one case of a de novo complex chromosomal rearrangement (CCR) involving three different chromosomes,1, 5 and 12. Two pregnancies of the female carrier over three years resulted in two spontaneous abortions. Initial cytogenetic analysis of her peripheral lymphocyte by G banding suspected a karyotype 46,XX,t(1;5;12)(1pter →1q25::12q24→12qter;5qter→ 5p11::1q25→1qter;12pter →12q24::5p11→5pter). Fluorescense in -situ hybridization (FISH) was used to confirm the karyotype 46,XX,t(1;5;12)(1pter →1q23::12q22→12qter;5qter→5p11::1q25→1qter;12pter→12q22::1q23→1q25::5p11→5pter). Seven years later she was pregnant again and refused to have prenatal diagnosis. The fetus is normal both in phenotype and karyotype。Comparing previously reported female CCR carriers with the case, we conclude that female CCR carriers may not always present spontaneous abortion or have offspring with congenital malformation and can have chance to get a healthy child.

关键词　复杂染色体易位　女性携带者　健康女儿Key words　complex chromosomal rearrangement　female carrier　healthy girl

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Abstract

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