The 102-year old woman with translocation (7;12) and infertility in anamnesis

Alina WOJDA1, Danuta WOLNIK-BRZOZOWSKA2, Maria LUBKA1, Małgorzata MOSSAKOWSKA3, Michał WITT1,3

1 Institute of Human Genetics, Polish Academy of Sciences, Poznań, Poland  
2 Department of Medical Genetics, Medical University, Poznań, Poland  
3 International Institute of Molecular and Cell Biology, Warszawa, Poland

Abstract. In this study we present a 102-year old woman carrying a (7;12)(q11.3;q14) translocation. A woman displays a normal phenotype and infertility in anamnesis. This is the first report linking t(7;12) and infertility.

Key words: centenarian, chromosomal aberrations, infertility.

Balanced carriers of a reciprocal translocation are at an increased risk of chromosomally unbalanced offspring, spontaneous abortions or infertility. During meiosis, the segregation of the translocated chromosomes and their normal homologs produces unbalanced gametes and an unbalanced zygote or gametogenic breakdown, fertilization failure or early zygote development arrest and miscarriage (GOLDMAN at al. 1992, VAN DER VEN et al. 1998, FARAUT at al. 2000).

A 102-year old woman presented in this report was referred for medical examination within the project of the medical/genetic investigation of Polish centenarians. She was phenotypically normal and her height was 165 cm. She had never been hospitalized but had fertility problems. The woman studied was married twice but she never became pregnant. The presented woman had two sisters and four brothers, of whom all had healthy offspring. Due to the fact that the subject died before the study was completed, a family history interview was conducted with the centenarian’s cousin.

Received: July 10, 2003. Accepted: July 17, 2003.
Correspondence: A. WOJDA, Institute of Human Genetics, Polish Academy of Sciences, ul. Strzeszyńska 32, 60-479 Poznań, Poland, e-mail: wojdal@man.poznan.pl
Cytogenetic investigation was performed on peripheral blood lymphocytes using the G banding technique. The karyotype was analysed in at least 20 metaphases. It revealed a balanced chromosomal translocation between chromosome 7 and chromosome 12. The karyotype was 46,XX,t(7;12)(q11.3;q14).

Although in older individuals up to 10% of somatic cells are affected by chromosomal aberrations (MAURER et al. 2003), this particular one should not be counted.

Figure 1. Facial appearance of the patient in the age of 28 (a), and about 60 (b)

Figure 2. Partial G banded karyotype showing the translocation (7;12)(q11.3;q14). Brackets show translocated segments of chromosomes.

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as the effect of cellular senescence since specific translocations involving chromosome 7 represent in fact an exception to the frequently observed age-related increase of structural aberrations: they are more frequent in newborns than in adults (PRIEUR et al. 1988). Moreover, the aberration was present in all the studied cells. Familial cytogenetic investigation was not feasible but on the basis of anamnesis (a lack of miscarriage, abnormal children or infertility in the family) it can be assumed that this translocation can either be passed from generation to generation without any effect on fertility or, as a de novo mutational event, causes infertility in the affected individual. The latter is the most likely mechanism causing infertility in the woman presented in this report. Literature search shows that no data linking t(7;12) with infertility has been reported so far.

Acknowledgement. This work was supported within grant no. PBZ-KBN-022/PO5/1999 “Genetic and environmental factors of longevity” of the State Committee for Scientific Research (KBN) coordinated by the International Institute of Molecular and Cell Biology in Warsaw.

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