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Citation	北海道大學理學部紀要, 16(1), 38-40
Issue Date	1966-12
Doc URL	http://hdl.handle.net/2115/27422
Type	bulletin (article)
File Information	16(1)_P38-40.pdf



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A Human Female with a D/G Chromosome Translocation¹⁾

By

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(With 2 Text-figures)

While working with chromosome aberrations in patients with infectious hepatitis, the present author had an opportunity to study a chromosome abnormality involving a D/G translocation in a female patient. In the present paper, the author wishes to present in some detail the results of the chromosome analysis undertaken in that subject with some remarks on the nature and origin of the abnormal chromosome.

It is the author's pleasant duty to express here his sincere gratitude to Professor Sajiro Makino who read through this manuscript, for his special direction with invaluable suggestion and advice.

Materials and Methods: The propositus who was hospitalized due to infectious hepatitis was an unmarried nurse, aged 26 years. In general appearance, she was phenotypically and mentally normal. The pedigree relative to her is presented in Figure 1. The members of her family were all mentally and physically normal: there was no woman who had spontaneous abortions. Chromosomal study was carried out on the propositus based on short term cultures of leucocytes derived from the peripheral blood. Chromosome slides were made according to the method of Moorhead *et al.* (1960) with a slight modification.

Results and Remarks

On the basis of chromosome counts of one hundred metaphasic cells, it was found that the propositus had 45 chromosomes. Detailed chromosome analysis revealed that this woman was characterized by a D/G translocation. In other words, two chromosomes were lacking, one from D group and the other from G group, with an extra element in group 6-X-12 in all cells studied (Fig. 2). One of chromosomes of group 6-X-12 was remarkable by carrying a secondary constriction. Apparently the said element corresponds to a translocated chromosome.

1) Contribution No. 761 from Zoological Institute, Faculty of Science, Hokkaido University, Sapporo, Japan.

Jour. Fac. Sci. Hokkaido Univ. Ser. VI, Zool. 16, 1966.

Theoretically, there should be present a centric fragment produced by reciprocal translocation, but no such element could be actually identified in any cell under study. In a study of chromosomes of a mongol with translocation, Barnicot *et al.*

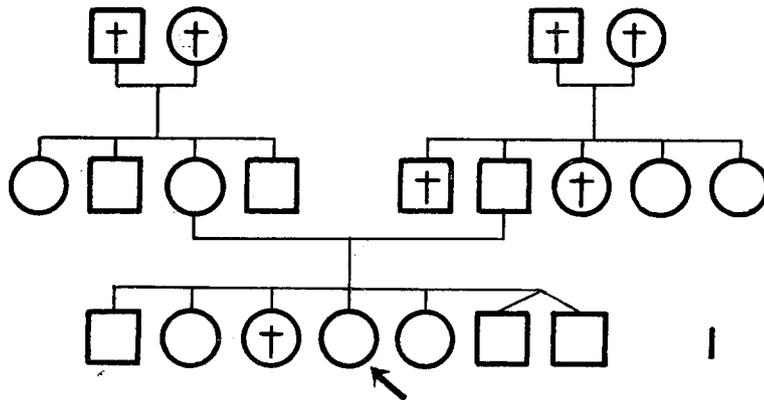


Fig. 1. Pedigree of the propositus. An arrow indicates the propositus.

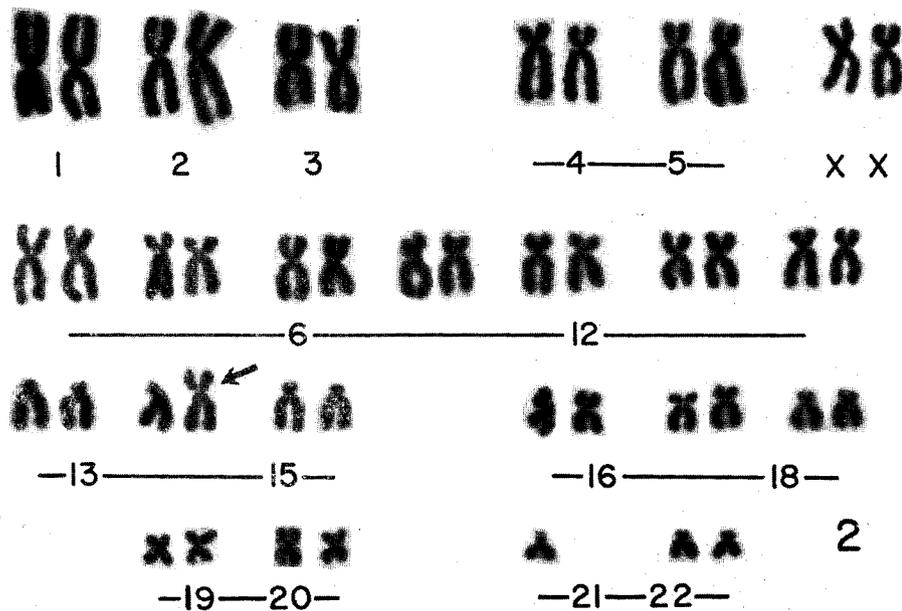


Fig. 2. Karyotype analysis of a cultured leucocyte from the propositus showing D/G translocation. The chromosome with an arrow seems to be the translocated chromosomes.

(1963) described that the unusual elongation of the centromere region in a D/G translocated chromosome might be caused by translocation. Breg *et al.* (1962) observed one chromosome in group 6-X-12 with a secondary constriction close to the centromere, and stated that it was the chromosome resulted from a translocation between the chromosomes no. 15 and no 21. The information on the translocation as cited above seems to be significant for the interpretation of the possible origin of the D/G translocation occurring in the present case. Recently, Makino *et al.* (1965) reported a female patient hospitalized due to serum hepatitis and one of her two sons who showed a chromosomally balanced B/C reciprocal translocation, all being phenotypically normal. Based on the fact that the propositus cast two successive spontaneous abortions in the past, it was presumed that the B/C translocation may be associated with a cause of the abortion.

Summary

A 26-year-old female patient hospitalized due to infectious hepatitis was found to carry a D/G translocation. The propositus and her relatives are all normal both mentally and physically.

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