A SURVEY OF A FAMILIAL TRANSMISSION OF AN ANOMALOUS AUTOSOME IN GROUP 13-15*

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Abstract. A male child hospitalized due to undescended testes (cryptorchism) was found to possess an abnormal autosome with an unusually elongated short arm in group 13-15. A familial chromosome investigation undertaken in 14 persons related to the propositus in his paternal line and in his mother revealed that his clinically normal father, grandfather, 2 aunts and a female cousin carried the same aberrant autosome. It is evident that a carrier of the abnormal chromosome is the grandfather, that the anomalous element was transmitted, irrespective of sex, from the parents either one of whom carried the aberrant one, and that the particular autosomal abnormality is not always associated with specific phenotypic anomaly. A possible origin of the aberrant autosome is discussed.

Introduction

While the senior author was conducting a chromosome survey since 1959 on normal and abnormal human subjects in the Japanese population, it was found by chance that a child with undescended testes (cryptorchism) and his clinically normal father carried a previously undescribed abnormal autosome with an unusually enlarged short arm occurring in group 13-15, whereas the mother showed no such aberration. This particular abnormality has been given a preliminary description by the senior author and his colleagues (KATO, OMURA, TABATA and MAKINO, 1965). The results are suggestive that the anomalous autosome is apparently inheritable through paternal transmission.

In order to obtain further information on the familial transmission of this particular abnormality, a chromosomal survey has been continued by the authors among the relatives of the paternal family of the propositus characterized by an unusual autosome (group 13-15).

The present paper reports the results of this survey as concerns the transmission pattern of the abnormal chromosome, its origin and nature, and its genetic significance in relation to a possible incidence of certain developmental anomalies.

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The senior author (S. M.) wishes to dedicate this paper to Dr. JAKOB SEILER on the occasion of his 80th birthday, May 16, 1966.

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Materials and Methods

Clinical records of the propositus with undescended testes have already been described in some detail in a preliminary paper (Kato, Omura, Tabata and Makiyo, 1965). Brief clinical features are given: The propositus is a 18-month-old boy who is a first and only child born to parents who are physically and mentally normal. The father's and mother's ages at the birth of the propositus were 27 and 25 years, respectively. Clinically the patient was found to have undescended testes, though not by operation, together with congenital dislocation of the hip. His mental and some other physical developmental features appeared normal for his age, showing no sign of Down's syndrome.

It became evident by personal interview with the patient's parents that all paternal living sibls are married and each has two or three children, respectively, and that no congenital disease was noted in the relatives other than the propositus here considered.

The paternal pedigree of the patient is presented in Figure 1. Sixteen persons from paternal siblings including the mother of the propositus, ranging in age from 18 months to 77 years, were subjected to chromosomal study: they are the propositus, his father, mother, grandparents, 2 uncles, 3 aunts, and 6 cousins (Fig. 1).

Chromosome studies were exclusively carried out on the basis of short-term leucocyte cultures established by venous blood from 16 individuals as mentioned above. Chromosome slides were made according to the air-drying method of Moorhead, Nowell, Mellman, Batts and Hungerford, (1960) after a slight modification.

Results

The following Table provides the results of chromosome counts in 15 persons related to the propositus from the paternal pedigree and in his mother. The data presented in the Table indicate that every one under