

## PANEL II: HUMAN CHROMOSOMAL DISORDERS

Reporter - TOM MARRIE

This panel was chaired by Dr. Malcolm Ferguson Smith of Glasgow University. The panelists dealt with what they considered five growing points in this field:

- (1) Chromosomal Breakage
- (2) The XXY Syndrome
- (3) Monosomy G
- (4) Polyploidy
- (5) Meiotic Analysis

Dr. James German from the New York Blood Center considered Chromosomal Breakage under three headings, consequences, known causes and possible and probable implications. He went on to conclude that in the childbearing age causes of chromosomal breakage should be avoided.

The XXY Syndrome was considered by Dr. Philip Welch of Dalhousie. He noted that these individuals are tall, aggressive, and are subnormal intellectually. They have a performance I.Q. that is greater than their verbal I.Q. There is also a slight lengthening of the PR interval in the E.K.G. The first cases of this syndrome were discovered in prisons, and Dr. Welch answered a question that had been asked by many - Does the possession of an extra "y" chromosome predispose to crime? The answer is no. These individuals come from families where there is a high incidence of broken homes. Interestingly enough most of the crimes are against people instead of against property. Dr. Jacques Gagnon of the Hospital Ste.

Justine in Montreal told of his work on Monosomy G a condition in which there are only 45 chromosomes in some cells. Up until the present it was believed that the missing chromosome was chromosome number 21. For this reason the syndrome was called the "Antimongoloid" syndrome. By autoradiography Dr. Gagnon showed in his case that it is really chromosome number 22 that is missing.

The subject of Polyploidy was considered by Dr. David Carr of McMaster University. He found that in women who became pregnant six months after ceasing to take the contraceptive pill the incidence of polyploidy was greatly increased. However, this condition is incompatible with life and all the conceptuses were aborted. The incidence of trisomy was the same as in a control group. He concluded that there is something different about women who have stopped taking the pill.

Dr. Malcolm Ferguson-Smith considered Meiotic analysis as one of the methods of detecting smaller aberrations in chromosomes. The smallest deletion that is visible with the microscope contains about 30,000 genes. Material for Meiotic analysis can be obtained very easily in the male by biopsing the testis. In the female a biopsy of the ovary is required and to increase the number of useful ova gonadotrophins should be given before the procedure.

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