For General Practitioners:

CHROMOSOMES IN HEALTH AND DISEASE*

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Introduction

Physicians have long been regarded by their patients as advisers—more so, a general practitioner, because he occupies a strategic position in the family group. A family physician has the advantages of caring for more than two generations at a time, which gives him the opportunity to observe the inheritance of various diseases, and to assume the responsibility of genetic counselling. The fear of having another affected child may cause parents to avoid having a child when in fact they could have been assured that the risk was insignificant. It is rather fortunate that genetic principles required for counselling are often not very complicated. Through a detailed family history and a thorough investigation of exceptional cases, the general practitioner can contribute much to the new breakthrough in genetics and thus play an important role in preventive medicine.

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carry the genes which are the carriers of various traits. In the simplest form of inheritance only one site on the chromosome carries one particular gene which produces a trait. As chromosomes occur in pairs there are two genes for each character. A pair of chromosomes can be thought of as two groups of genes. The gene for a particular character occupies a specific site on a chromosome. If both chromosomes of a pair contain the same gene the organism is said to be homozygous with respect to that character. If different genes are present the organism is heterozygous.

The chromosomes can be cultured from bone marrow, fibroblasts from fascia lata or skin, leucocytes from peripheral blood, as well as from other tissues. Peripheral blood is the most convenient, easy to collect and need only be cultured for three days. An adequate number of dividing cells for good chromosome preparation is ensured by the addition to the culture of either colchicine or the other related alkaloid, demecolcin. This material arrests cell division at the metaphase stage when the chromosomes are better visualized and are most suitable for study.

The chromosomes are classified into various groups by their relative total length and position of the centromere. Group A includes chromosome pairs 1, 2 and 3; Group B comprises chromosome pairs 4 and 5; Group C comprises chromosome pairs 6 to 12 and also the X(sex) chromosome (two in the female and one in the male); Group D comprises pairs 13-15; Group E includes pairs 16, 17 and 18; Group F, pairs 19 and 20; Group G, pairs 21 and 22. The male Y chromosome is very much like the ‘G’ chromosomes, and is included in this Group.

In the human female, the sex chromosomes (usually called X chromosomes) are similar; in the male, a chromosome called the Y chromosome is paired with an X chromosome. Testicular differentiation is directed by genetic information contained in the Y chromosome.

The number of X chromosomes in an individual can be easily studied by examining stained scrapings of the buccal mucosa. This is called nuclear sexing, on the basis of finding a ‘Barr’ body or sex chromatin in females and the absence of it in males. It is formed by one of the two X chromosomes in females, hence it is characteristically present in the normal female and absent in the normal male. This sex chromatin is located on the inner surface of the nuclear membrane and it consists of a chromatin mass of one micron in diameter. In general the number of Barr bodies is one less than the number of X chromosomes. The discovery of the sex chromatin mass has made possible the recognition of some types of hermaphroditism and other anomalies of the sex chromosomes in early infancy, by a simple method.

Chromosomal Disorders

Several clinical syndromes with autosomal or sex chromosomal anomalies are now recognized.

*Down’s syndrome or mongolism.* The main features are the oblique palpebral fissure, short broad hands, short crooked incurved fifth finger, hyperflexibility, epicanthus, furrowed tongue, flat occiput, narrow high arched palate, mental retardation, irre-