

SEX CHROMATIN AND LYONIZATION

RAMSADAY COLLEGE

LOAKNATH GHOSH

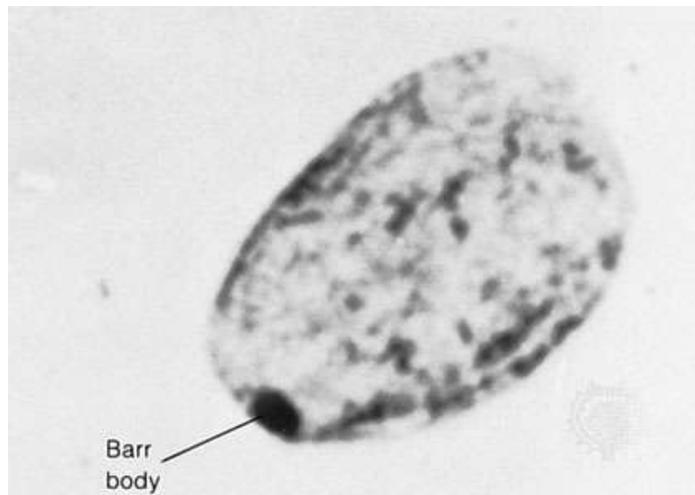
The term 'sex chromatin' denotes a body in the nuclei of cells, which indicates the sex of the organism from which the cell originated. The sex chromatin body is present in the nucleus only during interphase and disappears during mitosis.

The discovery of sex chromatin is generally placed in 1949, when Barr and Bertram, in London, Ontario, described a morphological distinction between neurones of male and female cats. The sex difference consisted of a small body which was present in the nuclei of nerve cells from female cats but was absent in those of males. In 1952, Graham and Barr were able to show that in the cat sex chromatin could be demonstrated in tissues other than neurones. Sex chromatin was found in all tissues (except in liver and pancreatic acinar cells). In 1953, the sex difference found in human and it's became a part of human genetics.

In 1959, a striking correlation was revealed between the sex chromatin status and the number of X chromosomes in an individual. The presence of sex chromatin was found to be associated with the presence of two X chromosomes. The term 'sex chromatin' is now called '**Barr bodies**'.

BARR BODIES

A Barr body is a small well-defined body which stains intensely with nuclear dyes. It is present in a large proportion of nuclei of female origin and absent in male nuclei. The size of a Barr body is about 1μ in diameter.



Barr bodies are most commonly situated at the periphery of the nucleus

NUMBERS OF BARR BODIES PER NUCLEUS

In general, the maximum number of Barrbodies per nucleus in any organism or tissue is either **0** or **1**, corresponding to a karyotype containing one or two X chromosomes respectively. However, cells with multiple Barr bodies may be countered, and this in variably means that more than two X chromosomes are present in the karyotype. The excess of X chromosomes may be due either to aneuploidy (e.g. if three X chromosomes are present in a diploid nucleus) or to polyploidy (e.g. as a result of doubling a female chromosome complement, resulting in a tetraploid nucleus with four X chromosomes). The Y chromosome appears to have no effect on the formation of Barr bodies, even though it results in a male phenotype.

Barr body was formed from a single X chromosome. One chromosome in the female appeared to be more condensed than the rest and that such a condensed chromosome was not seen in the male. Tetraploid cells in the female appeared to have two such chromosomes.

THEORETICAL CONSIDERATIONS CONCERNING SEX CHROMATIN

The inactive X hypothesis in its wider form was advanced and developed by Lyon (1961, 1962 and 1963).

The Lyon hypothesis consists of 3 parts.

First, it postulates that the genes on the sex chromatin forming X chromosome are inactive.

Secondly, that the decision as to which of a pair of X chromosomes is to be inactivated is made early in embryonic life and that, once the decision is made, the descendant so feach X chromosome will be like the parent chromosome.

Thirdly, that the original inactivation in each embryonic cell occurs at random, so that in some cells a paternal and in others a maternal X chromosome will be inactivated.

DOSAGE COMPENSATION

The term 'dosage compensation' was introduced by Muller (1932) to describe the fact that in *Drosophila* the effect of genes borne on the X chromosome is about the same in males where they occur in single dose, and in females where they are present in double dose. Muller's explanation was that the genes on the X chromosome were balanced in such a way that the effects of dosage were cancelled.

Since the effect of the theory would be that animals of both sexes have only a single dose of sex linked genes, it is thought to provide the basis for dosage compensation in mammals (Lyon, 1963).

Lyonization is commonly known as X-inactivation. In mammals, males receive one copy of the X chromosome while females receive two copies. To prevent female cells from having twice as many gene products from the X chromosomes as males, one copy of the X chromosome in each female cell is inactivated.

EXAMPLE

A classic example of X-inactivation is seen in cats. If a female cat is heterozygous for black and tan alleles of a coat color gene found on the X, she will inactivate her two Xs (and thus, the two alleles of the coat color gene) at random in different cells during development.

The result of is a tortoiseshell coat pattern, made up of alternating patches of black and tan fur. The black patches come from groups of cells in which the X with the black allele is active, while the tan patches come from cells in which the X with the tan allele is active.