9. The genetics of sex

My name is Dorota Kołodziejczyk. I'm a lecturer at Siedlce University of Natural Sciences and Humanities in the Institute of Animal Sciences and Fisheries. It's my pleasure to present to you the topic of The genetics of sex.
The essence of the science of heredity is to study the mechanisms by means of which a developmental plan is imposed on the offspring, leading to the formation of an organism with general structural features and functions characteristic of all individuals of this species, and similar in specific features to its parents.
The concept of the origin of a trait, later called a gene, was introduced by Mendel, while Strasburger and Meizel made a significant contribution to the discovery and description of chromosomes and the understanding of the mechanisms of cell division - mitosis and meiosis.
At the beginning of the 20th century, geneticist Thomas Morgan developed the chromosomal theory of inheritance, proving that chromosomes are the carrier of genetic information.
They are nucloproteins, and their essential component is deoxyribonucleic acid (DNA), which is a hereditary substance.
Chromosomes are divided into two basic groups,
namely:

- autosomes,
- heterosomes, otherwise known as sex chromosomes.

Sex chromosomes are formed from a pair of autosomal chromosomes, which through progressive specialization have accumulated a large number of genes responsible for gender determination.
Sex-determining genes are not only localized in both heterosomes and autosomes. Moreover, in different groups of animals, the mutual importance of hetero- and autosomes in genetic sex determination is not the same.

GENETIC SEX DETERMINATION IN MAMMALS
In mammals there is a genetic sex determination mechanism - XX/XY, with a few exceptions among the order of marsupials. The heterogametic sex, i.e. the one that produces two types of gametes, is male (XY) in mammals, while females are the homogametic sex (XX).
The decisive role in sex determination in mammals is played by the male Y chromosome, which exerts an influence on the initially female-determined embryo. This is due to the SRY (sex determining region Y) gene located on this chromosome. It is this gene that initiates and conditions the transformations that result in the development of primary sex characteristics - testicles, determining male sex.

Male Y chromosome
The absence of the SRY gene on the Y chromosome leads to the creation of an individual with a female phenotype despite the presence of this Y chromosome.
In the gonads (testes and ovaries), meiotic cell division occurs, resulting in the formation of gametes. In this process, the sex chromosomes are randomly sorted and all eggs carrying an X chromosome are produced in the female, and the X or Y chromosome goes to the sperm.
In order for female cells not to make twice as many proteins encoded on the sex chromosomes, one must be turned off in some way. This happens in early embryonic development, where one of the X chromosomes is randomly inactivated in cells and becomes visible as a lump of sex chromatin (Barr body).

GENETIC SEX DETERMINATION IN BIRDS

Unlike mammals, birds are characterized by the genetic mechanism of sex determination - ZZ/ZW. The heterogametic sex in birds is the female (ZW) and the males are the homogametic sex (ZZ).

It is interesting that in birds, as in mammals, sex chromatin was observed in females, which indicates that the sex chromatin clump is not formed as a result of the fusion of two homologous sex chromosomes.

It has been shown that in birds the ovary is the dominant gonad and if the ovary is implanted next to the testis, it transforms into an ovary (Boczkowski, 1983).

According to other authors (Nowak, ed., 2015), the role of the Z and W chromosomes is not yet fully resolved. In birds, the SRY gene responsible for the formation of the male phenotype was not found. Scientists believe that the level of the DMRT1 gene product mapped on the Z chromosome is responsible for determining the bird's sex.

GENETIC SEX DETERMINATION IN INSECTS

Research on genetic sex determination in insects has become the basis for examining the genetic mechanisms of sex determination.

The turning point was Jan Dzierzon's discovery of the phenomenon of parthenogenesis (1845). A Polish priest and beekeeper stated and proved that the queen bee lays two types of eggs: fertilized and unfertilized.

From the latter, males are formed - drones. Fertilized eggs, on the other hand, hatch into workers or queens (depending on the diet - if it is poor in protein, it causes sexual underdevelopment of workers).

Egg cell n + no sperm = n male;
Egg cell n + sperm = 2n female.

Based on research on Drosophila, it was found that sex depends on the ratio of the number of X chromosomes to the number of haploid autosomal suits, with X chromosomes affecting the development of females and autosomes affecting male development. The presence of the Y chromosome does not determine sex, but it is important for the full development of testicular function.

SEX CHROMOSOMES AND AUTOSOMS	RATIO X:A	GENDER DEVELOPMENT
XY + 2A (diploid)	0,5	male
X + 2A (monosomic)	0,5	infertile male
XX + 2A (diploid)	1,0	female
XXX + 2A (trisomic)	1,5	super female
XXX + 3A (triploid)	1,0	female
XYY + 3A (triploid)	0,33	male
XXY + 3A (triploid)	0,67	intersex
XXXX + 4A (tetraploid)	1,0	female
XXXY + 4A (tetraploid)	0,75	intersex

GENETIC SEX DETERMINATION IN FISH

Fish show the greatest variety of sexual differentiation. Unlike other vertebrates, hermaphroditism (bisexuality) is a physiological condition in fish. There are 5 types of hermaphroditism in this group of animals:

- Synchronous hermaphroditism – a state in which an individual is capable of functioning as both male and female simultaneously;
- Alternating functional hermaphroditism – that is, an individual functions once as female, once as male;
- Protandric hermaphroditism – a state in which an individual functions first as male, then as female;
- Protogynoid hermaphroditism – a state in which an individual functions first as female and then as male;
- Residual hermaphroditism – means the existence of hermaphroditic features in an individual functioning only as male or female..
  (Boczkowski, 1983)

GENDER-LINKED CHARACTERISTICS

Many traits in humans and animals are determined by genes located on the sex chromosomes, hence they are called sex-linked traits.

Some of them are:

- hemophilia (in humans and dogs)
- leg twisting (in pigs),
- a lethal gene, causing the absence of teeth and the death of male fetuses (in cattle),
- black and yellow coloration of the coat (in cats),
- hawklike plumage (in hens),
- polledness (in cattle),
- fruit fly eye color.

GENDER-RELATED CHARACTERISTICS

Sex-linked traits are those that are determined by genes located in autosomes, but their phenotypic expression depends on the sex of the individual.

For example, red or mahogany color in cattle is determined by the Mm gene pair. Dominant homozygotes - MM, regardless of sex, are mahogany, homozygous recessive - mm, are red, both males and females, while the color of heterozygotes - Mm depends on sex - males are mahogany and females are red (Świderek, 2015).

GENDER-RESTRICTED FEATURES

Sex-restricted traits are traits that occur in only one sex, even though the genes for that trait are passed onto offspring by both parents.

The genes determining these traits can be located on both autosomes and sex chromosomes.

Some of them are:

- milk secretion in mammals,
- egg-laying in birds,
- cryptorchidism in males,
- impotence of Friesian bulls
- fertility disorders of white shorthorn heifers (Maciejowski i Zięba, 1982; Świderek, 2015).

SUMMARY
Understanding the molecular basis of sex determination plays an important role in breeding.
Early assessment of the sex of the embryos makes it possible to obtain offspring of a specific, desired sex.
Embryos can be obtained by in vitro fertilization, cloning, or from superovulatory donor females and transferred to recipient females.
A useful method of determining the sex of embryos is the detection of the gene sequence of DNA isolated from the cells of the male embryo. This is done on the basis of PCR amplification of a selected DNA fragment and electrophoresis of the obtained products.

Thank you for your attention!