MENDELIAN INHERITANCE IN HUMANS

1. Hello, welcome to the second lecture of the Module entitled Mendelian genetics. In the previous lecture we were showing Mendel's discoveries on how segregation and independent assortment work in pea plants. Now, we would like to turn our attention to humans.

2. As you remember, Mendel picked seven visible pea characteristics that were easy to distinguish. One of them was plant height. Mendel classified his plants into two types: tall and dwarf. They were so distinct that no measurement was necessary to differentiate between them.

3. In humans, you may also try to categorize individuals according to their height as tall or short

4. … but it is difficult to set up only two phenotypes, as height in humans is an example of a phenotype with continuous variation – a distribution of phenotypic characters that is distributed from one extreme to another in an overlapping, or continuous fashion. It is so, because height in humans is not controlled by a single gene pair, as it is in pea plants. In humans it is a complex trait determined by several gene pairs and environmental interactions.

5. As a matter of fact, most of our visible features, like for example eye or skin colour, do not have a simple one-locus, two-allele, dominant vs. recessive method of inheritance. After Mendel's work was rediscovered, some scientists believed even that inheritance of traits in humans might not work the same way as it did in plants and other animals.

6. However, in 1902, an English physician, sir Archibald Garrod described the first recessive human trait – alkaptonuria. It is a rare, but not serious familial disease. Individuals afflicted with this disorder cannot metabolize alkapton, also known as homogentisic acid. As a result it accumulates in cells and tissues. It is also excreted in the urine which turns black after it is exposed to the air. By studying the disorder's pattern of inheritance, Garrod concluded that alkaptonuria was inherited as a simple recessive trait. On the basis of this conclusion, he hypothesized that hereditary information controls chemical reactions in the body, and that the inherited disorders he studied are the result of alternative modes of metabolism. If you are interested to read Garrod's paper – you will find it in the supplementary materials for this Module.

7. Purely Mendelian traits are a tiny minority of all our traits and they mostly refer to "negative" phenotypes, disorders which are interchangeably called Mendelian, single-gene, or monogenic diseases. Some of them will be presented in Module 3 of our course.
You may find all known human Mendelian traits in a comprehensive catalogue of human genes and genetic disorders - Online Mendelian Inheritance in Man (OMIM), freely available via the internet. In the fifth Module of our course you will learn how to navigate it.

8. Talking about Mendelian inheritance in humans, it is good to be aware, that there still exist some common misconceptions about human genetics and heredity. For example, you might have heard that rolling your tongue is dominant to non-rolling. That's because this trait was originally described as fitting a simple genetic model. Well, it doesn't. Rolling our tongue it is not a simple two-allele character and neither are many other traits often used to teach the basics of genetics. Please, spare some time to visit an excellent website on the Myths of Human Genetics maintained by John H. McDonald of the University of Delaware. He debunks many of

the common examples of hereditary traits and discusses some good reasons why we shouldn't be teaching them.