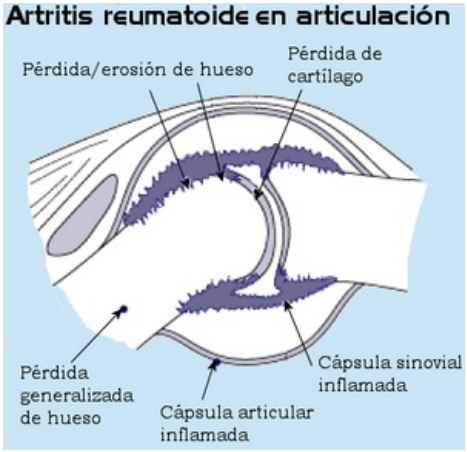
Autosomal recessive inheritance

Autosomal recessive inheritance *(AR)* refers to genes located on non-sex chromosomes - **autosomes**. We trace the transmission of a trait at a conditional recessive by an allele. Phenotypically, the monitored trait is **manifested'** only in **recessive homozygotes'** (*aa*). Heterozygotesi (*Aa*) are **without clinical manifestation**, they cannot be distinguished phenotypically from dominant homozygotes (*AA*). In the case of a *compound heterozygote*, i.e. a heterozygote for 2 different mutations (both alleles are mutated, but each differently), we get a recessive homozygote ("aa") phenotypically.



An example of a family tree with an autosomal recessive trait

Genealogical characteristics

🔎 *For more information see Genealogy.*

In AR, **both sexes are affected equally often*. Typically, it is a horizontal type of inheritance'*** - the parents are usually healthy (heterozygotes, carriers), the disease manifests itself in the offspring (occurrence of the disease "every generation"). In general, the probability of the manifestation of the disease increases with ""consanguineous marriages".

Risk calculation

When crossing two heterozygotes (*Aa*) there is a quarter probability (*25%*) of the birth of an affected offspring (***aa'**). Among the healthy offspring, two thirds will be carriers - heterozygotes.*

	A	**a**
A	AA	Aa
a	Aa	**aa**

When a recessive homozygote (*aa*) is crossed with a heterozygote (*Aa*) there is a half (*50%**) probability of having an affected offspring (***aa'**).

	a	**a**
A	Aa	Aa
a	**aa**	**aa**

Deviations

Deviations from AR include:

1. **genetic heterogeneity (heterogeneity)**

- a given disease (with a similar or identical phenotypic manifestation) can be caused by a mutation of one of several possible genes, i.e. a patient with both affected alleles of one gene on chromosome 11 and a patient with two mutated alleles of another gene on chromosome 18 can have a clinically similar disease, but the offspring of these two patients is likely to be healthy because they will be heterozygous for both genes
  - albinism (due to different genes; the offspring of two recessive homozygotes can be pigmented in some cases)
  - deaf-mute (due to a number of different genes, not to be confused with polygenic inheritance; the offspring of two deaf-mutes can also hear normally, cave! there are also dominant forms of deaf-mute).

2. pseudo-dominance.

Examples

- Cystic fibrosis – frequency in the population 1: 2.500[1]
- Phenylketonuria - frequency in the population 1:10 000[1]
- Sickle cell disease
- Werdnig-Hoffmann disease (OMIM: 253300 (https://www.omim.org/entry/253300))[2]

Links

Related articles

- Autosomal dominant inheritance
- Gonosomal inheritance
  - Gonosomal dominant inheritance
  - Gonosomal recessive inheritance
- Panmixie

Exercising

- Diseases - learning about heredity

Reference

1.
2. LISSAUER, Tom – CLAYDEN, Graham. *Illustrated Textbook of Paediatrics.* 3. edition. Elsevier, 2007. 113 pp. ISBN 978-07234-3398-9.

Sources

- THOMPSON, James Scott – THOMPSON, Margaret Wilson – NUSSBAUM, Robert L. *Clinical genetics: Thompson & Thompson.* 6. edition. Triton, 2004. 426 pp. ISBN 80-7254-475-6.