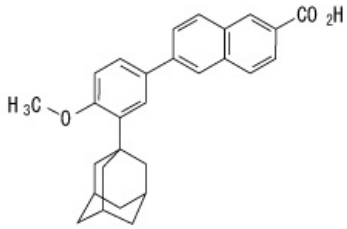
Autosomal dominant inheritance

Basic characteristics

- refers to genes located on non-sex chromosomes – **autosomes**
- we observe the transmission of a trait at a conditional dominant by an allele
- phenotypicwise, the monitored trait is manifested both in **heterozygotes** (*Aa*) and in **dominant homozygotes** (*AA*)
- in the case of incomplete dominance, heterozygotes (*Aa*) have **less severe *phenotypic manifestations than dominant homozygotes (AA), in which the respective disease manifests itself in a*** *very severe* form
- phenotypically healthy individuals (recessive homozygotes) do not transmit the mutation to subsequent generations



An example of a family tree with an autosomal dominantly inherited trait

Genealogical characteristics

🔎 *For more information see Genealogy.*

- '*both sexes are affected equally often*
- typically it is **vertical type of inheritance** - at least one parent is affected (most often heterozygous), the disease occurs in practically every generation

Risk calculation

- when two heterozygotes (*Aa*) are crossed, there is a three-quarters probability (*75%*) of the birth of an affected offspring (in 25% of cases, the affected individual is a dominant homozygous AA)

	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 the birth of an affected offspring (*Aa*)

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

Deviations

- sporadic cases - mutation *de novo* (new mutation) - common for example in achondroplasia
- incomplete penetrance (the allele manifests phenotypically in fewer carriers than we would expect)
- variable expressivity (variable degree of character manifestation)
- a consequence of the external environment or other genes
- late-onset disease – polycystic kidney disease, Huntington's chorea
- mosaicism of germ cells
- nonpaternity

Examples

- Achondroplasia
- Apert syndrome
- Brachydactyly
- Familial hypercholesterolemia - frequency in the population 1:500
- Huntington's chorea - frequency in the population 1:10 000-20 000
- Marfan syndrome
- Myotonic dystrophy – myotonia, muscular dystrophy, cataract, hypogonadism, frontal baldness and changes

EKG[1]

- Neurofibromatosis – frequency in the population 1:3 500
- Osteogenesis imperfecta
- Polycystic kidney disease - adult type, frequency in the population 1:1 000

- Polydactyly

Links

related articles

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

Exercising

- Diseases - learning about heredity

Reference

1. ADAMČOVÁ, Hana. *Neurology 2005.* Trendy v medicíně edition. Triton, 2005. 260 pp. ISBN 80-7254-613-9.

References

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