

Phenocopy non-inherited change of phenotype imitating the mutations, which is provoked by environmental factors. The graphical representation of pedigree (the family tree) with indication of relations and the presence of analyzable sign. The collection of information (interrogation and inspection) about presence (or absence) the sign (disease), which is analyzed and composition of legend about the relations of proband. The reason of second type is mutation of two genes of 6-pyruvoyltetrahydropterin synthase (PTS) caused PTS deficiency. Determination of analyzable disease's type (heritable disease, multifactorial (MFD) or phenocopy). Teratogens (different chemical or biological substances) disturb embryogenesis, but can't change a fetal genotype. Other type of congenital cataract is provoked by virus of German measles and is uninheritable. The reason of first (classical type) is the mutation of enzyme phenylalanine hydroxylase. Home task The genealogical method is the general and obligatory method of genetic consultation. In medicine it is uninheritable disease, which has the same clinical picture as monogenic heritable disease. Determine, who from family's members are the obligate and facultative heterozygote. One type of congenital cataract is heritable. The clinical pictures of both diseases are nearly identical. 5 .2.3.