PHENOCOPY

The term **phenocopy** refers to an individual whose phenotype has been altered by environment to a state that resembles a phenotype associated with another genotype. Phenocopy is not inheritable. A phenocopy is the development of a trait that looks like an inherited one, but is caused by a different genetic or environmental factor.

Phenocopy occurs when an individual presents a phenotype identical to that caused by genetic factors; however, in these patients, the phenotype is caused exclusively by environmental factors that mimic the behavior of the genetically produced one.

Examples of phenocopy

Breast cancer: A family history of breast cancer may be linked to a BRCA1 or BRCA2 mutation, but a woman who develops the disease might have a phenocopy, meaning she has breast cancer without having these specific genetic mutations.

Parkinson's disease: Familial Parkinson's disease is often caused by mutations in genes like SNCA, LRRK2, PRKN, and PINK1. However, a phenocopy occurs when a person has the disease but does not have mutations in these specific genes, likely due to other genetic or environmental factors.

Spinal muscular atrophy: This is a neurological disorder that can be a phenocopy, meaning different genetic anomalies can lead to a similar presentation.

Hereditary angioedema: This can be phenocopied by an acquired autoimmune form, where autoantibodies develop against the C1-inhibitor protein, even though the original condition was inherited.

Thyroid deficiency: A child with a genetically caused thyroid gland that does not produce enough thyroxine can show the normal phenotype after being treated with thyroxine hormones. This is an example of an environmental factor (treatment) leading to the normal phenotype, thus creating a phenocopy.