

# Genetic concept in psychiatry

**Inheritance of mental disorders** can be observed in more stable communities with limited migration (rural today, population dynamics are too high in cities).

## Linkage of hereditary factors and environmental influences

- Indicates the impossibility of studying genetic factors from the study of environmental influences - mental disorders are not always purely genetic, but arise under the influence of an exogenous factor.

### Pharmacogenetic interactions:

- When introducing **suxamethonium** for myorelaxation, someone was found to have an abnormal response (exaggerated muscle relaxation with necessary breath support, etc.), scientists discovered the enzyme **pseudocholinesterase** (atypical).
- **Hemolytic reaction** in persons with a hereditary defect *Glucose-6-phosphate dehydrogenase deficiency (G6PD)* - after certain drugs or **medicines** (antimalarial **primaquine**); hemolysis does not occur in persons without this mutation.

## Genealogy

- The main method of studying heredity – genealogy, has brought valuable empirical data, but has not yet solved the ways of transmission of some diseases;
- the main problem is with the definition of disorders - whether they are homogeneous or heterogeneous;
- these will rather be diseases with a similar clinical picture, but with different etiology;
- the main effort to create more homogeneous groups of probands;
- twin study.

The basis of the nosological categorization of mental disorders is still the *phenomenological description*.

## Galton's concept

- **Behavioral Sciences**, uses a biometric method for quantitative analysis;
- today will most likely not bring anything new, Mendel's laws of heredity will be decisive;
- enables detection of the participation of a genetic factor, but does not lead to an explanation of the genetic mechanism;
- abnormal function often caused by a single defect;
- on the other hand, the variability of a normal trait can rarely be attributed to a single allelic disorder, but is usually the interplay of several genes;
- in other words, normal behavior is controlled by many genes and environmental influences, but a major defect can also be caused by a malfunction in one of these components:
  - **intelligence** is, for example, a quantitative trait whose distribution in the population is represented by a Gaussian curve. According to this concept, control is given by many genes, a number of polygenes;
  - a single gene defect is often a major disorder such as *phenylketonuria*.

## The polygenic hypothesis

- Hereditary transmission through genes, the effect of which is mostly 'added up';
- disturbances in the field of *quantitative phenomena*;
- solves the nosological relationship of phenomena - e.g. the difference between malignant and benign schizophrenia - in the number of damaged "pathic genes";
- a good basis for interpreting the interplay of genetics and environment - the *threshold theory of schizophrenia*;
- this concept also seems to be exhausted.

## Qualitative concept

- Now moving mainly to concepts based on the qualitative concept;
- there are critical issues of defining homogeneous nosological groups and the probability of etiological heterogeneity;
- tries to affect subgroups of psychiatric patients who will be defined by a certain criterion (e.g. today's division of schizophrenia into positive and negative forms or examination of the connection between schizophrenia and the age of onset of symptoms).

## Analysis

- Logical consequence – shifting the emphasis of the concept to analysis – to gene detection, discovery of

- biological markers;
- for the time being, efforts to detect deviations in biochemical metabolism (phenylketonuria) prevail;
- contemporary psychiatry has almost given up hope of directly discovering a genetic defect.

Two main indirect research strategies are pursued:

1. **Searching for genetic markers:**
  - polymorphisms – genetic variants, different alleles at one genetic locus;
  - evolutionary processes;
  - study of polymorphisms – searching for mutant variants that may be related to mental illnesses;
  - e.g. relationship of disorders to MHC, blood groups, even at the molecular level.
2. **Gershon Genetic Vulnerability':**
  - chronic constitutional vulnerability of a disposed individual, even in clinical health;
  - her marker could reveal it to us, even if the person in question is without clinical symptoms;
  - the problem is that there may be other forms of the disease with similar clinical symptoms.

## Links

### Source

- BENEŠ, Jiří. *Studijní materiály* [online]. [cit. 16.02.2010]. <<http://jirben.wz.cz>>.