

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>>.