Some Considerations about Twin Zygosity and Concordance Determination in Cancer Research

R. P. Martynova

Studies on cancer in man and pure strain animals have shown that the malignant neoplasm, both in animals and in human beings, is not a single, genetically homogeneous disease: it consists of a lot of forms differing in localization, histological structure and etiopathogenesis. This circumstance is not always taken into account by the investigators in cancer research by the twin method, and especially in the determination of within-pair concordance/discordance, which is often a source of misunderstandings and faulty conclusions.

In order to establish a more objective criterion for estimating the real twin concordance in the studies of such diseases as cancer, we have worked out a quantitative index called Concordance Index:

$$CI = \frac{\frac{a}{n}}{\frac{a+b}{n}} \text{ 100,}$$

where: CI = Concordance Index %; a = number of twin pairs, MZ or DZ, which are fully concordant for the principal features; b = number of partially concordant twin pairs; n = total number of twin pairs, MZ or DZ.

The accuracy of twin zygosity determination acquires a particular importance in cases of low penetrance of the disease. In such cases, even a minor inaccuracy in the zygosity diagnosis may considerably influence the twins' CI, and always in the same direction: any mistake in the determination of twin zygosity always leads to an artificial diminution of the role of hereditary factors. Therefore, we consider that the questionnaire method of twin zygosity determination, as well as any other insufficiently objective method, should not be used in the genetic studies on cancer.

Considering the insufficient clinical examination of the twin propositi, it is suggested that the expression "twin method" be substituted by "clinical twin method" as if obliging the investigator to ensure a thorough clinical study of the twin propositi and their partners.

Prof. R. P. Martynova, Institut Citologij i Genetiki Sibirskogo Otdelenija Akademij Nauk SSSR, Novosibirsk 90, SSSR.