29 - Twin Studies

Twin Studies

© SPMM Course 12. Types of genetic studies Genetic methods can be classified into four paradigms

1. Basic genetic epidemiology: to quantify degree of familial aggregation and heritability estimates
2. Advanced genetic epidemiology: to explore the mechanism of action of genetic risk factors
3. Gene finding: to determine the genomic location and identity of offending genes
4. Molecular genetics: to trace biological pathways from DNA to disorder. Gene mapping refers to any strategy that permits finding the chromosomal location of one or more genes, often related to a disease. Genetic mapping of disease genes is a very useful method because it does not require any knowledge of a gene's function to find the chromosomal location initially. Once located then the identity of the disease gene could be dissected. Not all genetic studies are aimed at gene mapping; certain simpler designs are primarily aimed at demonstrating the presence or absence of a genetic influence in the aetiology of a disease or trait. These include family studies, twin studies, and adoption studies. Gene mapping studies involve linkage analysis, sib-pair analysis and to some extent allelic association studies. A. Classical genetic studies: Twin Studies Monozygotic (MZ, or "identical") twins are formed when an embryo is cleaved during early development. The result is two genetically identical embryos wherein 70% sharing even the same chorion. Dizygotic (DZ, or "fraternal") twins are the result of the fertilization of two different ova by two different sperm cells. DZ twins are genetically the same as siblings, sharing 50% of their genes. A pairwise concordance rate is estimated as the number of twin pairs who both have the disorder divided by the total number of pairs. However, where there has been systematic ascertainment, one can report a probandwise concordance rate, which is calculated as the number of affected twins divided by the total number of co-twins. This is possible if a twin register is maintained; it is also more useful method as this allows comparison of general population risk with the rate in co-twins of probands. Challenges in interpreting twin studies Monozygotes are often treated more closely than dizygotes as they look identical; so they share more environment than dizygotes. So a higher concordance may be due to higher environmental effect. Zygosity assignment done via anatomical similarity is far from perfect. Somatic mutations may occur in MZ twins after the cleavage event that forms them, causing "identical" twins to be at least somewhat different genetically. Chorionicity i.e. how many amnions and chorions are present for both foetuses determines shared uterine environment. Twin

studies assume that the risk of disorder is same in monozygotic and dizygotic pairs, and in singletons at the outset. This assumption holds good for most major psychiatric disorders, while it may not be the case for some physical disorders.

Revision #1
Created 2026-01-04 20:03:23 UTC by Omar Ayman
Updated 2026-01-04 20:03:23 UTC by Omar Ayman