57 - 14. Clinical genetics

14. Clinical genetics

© SPMM Course 14. Clinical genetics When an individual approaches a genetic clinic for genetic testing, 2 approaches can be employed. Direct testing: This is very much like any other lab test. A sample is tested for the presence of a certain genotype. Only one individual is tested, and the abnormality that is being sought is already known to have an association with the illness studied. Gene tracking: Many family members are tested to discover whether or not the suspected patient seeking the test has inherited the high-risk chromosome from a heterozygous parent. The test is based on Mendelian principles and seeks information about the segregation of a chromosome within a family. It can be used even if the exact genetic locus associated with a disease is unknown. . Prenatal identification: Prenatal test is the test of a fetus to identify a suspected genotype. It is often initiated on the basis of family history or maternal factors (e.g. older mothers at risk of Down's). Maternal serum screening to identify neural tube defects and Down's is offered routinely in many countries. In general, adult-onset genetic conditions are not usually tested prenatally. . Genetic counseling is routinely offered to individuals seeking genetic tests. The counseling service provides information about risks and probabilities before the test and also provides support (but not psychological) services after the testing. Within the NHS Regional Genetic Centres that incorporate cytogenetic, molecular and clinical genetic services operate and offer familial (carrier) testing, diagnostic and prenatal (presymptomatic) testing. . DNA banks provide secure, long-term storage for an individual's genetic material. While this is seen as beneficial for biomedical research the possibility of misuses has raised several ethical issues. .

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. DISCLAIMER: This material is developed from various revision notes assembled while preparing for MRCPsych exams. The content is periodically updated with excerpts from various published sources including peer-reviewed journals, websites, patient information leaflets and books. These sources are cited and acknowledged wherever possible; due to the structure of this material, acknowledgements have not been possible for every passage/fact that is common knowledge in psychiatry. We do not check the accuracy of drug related information using external sources; no part of these notes should be used as prescribing information.

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