

23 - 9. Cytogenetic techniques

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© SPMM Course □ VNTRs (variable number of tandem repeats). These polymorphisms are the result of varying numbers of repeats in a specific region of a chromosome. These Polymorphisms can be classified according to the length of polymorphic fragments; Short tandem repeat polymorphisms (STRPs) or microsatellites range in size from 2 to 6 bases. The minisatellites vary between 20 to 70 bases each. Microsatellites are currently preferred as genetic markers in disease mapping because they can be detected using the polymerase chain reaction. □ Polymorphisms arise out of mutations originally but are maintained in population due to number of factors such as founder effect, genetic drift and natural selection. □ Note that most polymorphisms occur in non-coding areas (introns) - as coding sequences or exons on mutation often produce disease phenotypes. □ Serotonin transporter polymorphisms are noted in promoter region, which is a non-coding part of DNA (5HTTLPR - 5HT transporter linked promoter region). 5HTTLPR can be of a short variant or long variant (length polymorphism). 55% of Europeans carry the long allele. In those with short variant, the serotonin transporter expression is low; short variant is speculated to be associated with higher incidence of affective disorders, neuroticism, anxiety and PTSD. But the evidence is inconclusive as most studies are case control design with significant heterogeneity. In an interesting study of environment-gene interaction, Caspi et al (2003) noted that individuals with one or two copies of the short allele of the 5-HT T promoter polymorphism exhibited more depressive symptoms, diagnosable depression, and suicidality in relation to stressful life events than individuals homozygous for the long allele. 9. Cytogenetic techniques

□ Blotting techniques □ Southern blotting is a widely used method for the detection of a specific sequence in DNA. This method was named after Dr. E. M. Southern who introduced this method in 1975. □ Western blotting is another widely used method for the detection of specific protein after electrophoresis. The sample is electrophoresed on a polyacrylamide gel, then, blotted to a membrane. The membrane is incubated with the antibody to the specific protein. □ Northern blotting is a detection method for a specific RNA after electrophoresis.

□ Polymerase chain reaction (PCR) Minute amounts of DNA can be amplified over a million times using an in vitro technique called polymerase chain reaction. Using this technique, minute amount of DNA such as those from buccal cell scrapings, blood spots, or single embryonic cells can be analysed. The DNA is amplified between two short single-stranded DNA fragments called oligonucleotide primers, which are complementary to the sequences at each end of the DNA of

interest. Hence the exact DNA sequence to be amplified needs to be

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