09 - Some deletion syndromes of psychiatric releva

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© SPMM Course is replaced by another purine or a pyrimidine replaced by another pyrimidine (e.g. A to G). It is called transversion if a purine is replaced by a pyrimidine or vice versa (e.g. A to T). According to the effect on triplet sequence, mutations could be frame shift or in-frame. In frame shift mutations, the deletion or insertion is not in multiples of three codons e.g. a segment of 5 bases deletion mutations. This leads to a shift in triplet reading frame with variable results. In frame, mutation refers to changes happening in multiples of 3 bases, with no disturbances in actual reading frame. According to the effect of a mutation on protein product, mutations could be silent, mis-sense or nonsense. A silent mutation causes no change in protein product – this is possible because a single amino acid is often coded by more than one triplet sequence. In a silent mutation one triplet sequence is replaced by a different sequence but without changing amino acid product. In mis-sense mutation, the new mutant codon specifies a different amino acid with variable effects on final protein product. For example, haemophilia, sickle cell anaemia. In non-sense mutation the new codon is UUA UGA or UAG, which signals ‘stop’ to the amino acid sequence resulting in nonfunctional protein. Point substitutions do not shift the reading frame; they often occur in non-coding regions and go unnoticed. Even at coding regions they are often silent or mis-sense mutations. Translocation refers to exchange of chunks of genetic materials from one chromosome to another. These are essentially mutations occurring at ‘larger’ dimensions. These are mostly reciprocal so one segment is exchanged for another segment among chromosomes. Robertsonian translocation is a non-reciprocal (i.e. unequal exchange) that results in a single fused chromosome from 2 acrocentric (non homologous) chromosomes. Following a Robertsonian translocation, the small 'p' arms are discarded, and a metacentric fusion chromosome results. Thus from 2 chromosomes a single chromosome is formed with no significant (only trivial) loss of genetic material. Hence, these are viable and ‘balanced’ within the individual in whom they occur. But when gametes are formed, only one of the two gametes can have the whole translocated

metacentric fusion chromosome, effectively resulting in monosomy (unbalanced translocation) for one gamete if fertilized and trisomy for the gamete with fused chromosome (extra load of genes now). This is one of the mechanisms for Down’s syndrome. Due to the mother being a carrier of such translocation, the recurrence rate of Down’s is extremely high in such cases compared to sporadic Down’s due to non-disjunction.

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