05 - Important trisomiesmonosomies

Important trisomies/monosomies

© SPMM Course Sometimes, non-disjunction can occur during mitosis immediately after two gametes have fused. This leads to the formation of two cell lineages, each with a different chromosomal make-up. This occurs more frequently with the sex chromosomes and results in a 'mosaic' individual. Mosaics exhibit milder malformations than those who carry complete aneuploidies. Important trisomies/monosomies Down’s syndrome: Trisomy 21 is the most common chromosomal disorder that occurs at a rate of 1:700 causing congenital mental retardation. Prominent findings are reduced maternal levels of αfetoprotein, increased β-hCG and increased nuchal fold thickness in fetal ultrasound. The child shows mental retardation, flat facial profile, prominent epicanthal folds, simian palmar crease, duodenal atresia, hypothyroidism and heart disease (most common malformation is septum primum–type ASD due to endocardial cushion defects). Alzheimer’s disease and leukaemia are common in affected adults who survive childhood difficulties. 95% of Down’s is attributed to meiotic nondisjunction of homologous chromosomes. This is associated with an advanced maternal age (rates are 1:1500 in women < 20 but 1:25 in women > 45). 4% of cases due to Robertsonian translocation and 1% of cases are attributed to Down’s mosaicism (no maternal age association is seen in these). The features of mosaic Down syndrome are milder but similar to the features of full Down syndrome. However, the clinical phenotype varies according to the level and distribution of trisomic cells. Thus, the affected individuals may range from completely normal to presenting the full expression of Down syndrome. Edwards’ syndrome is characterised by severe mental retardation and rocker bottom feet, low-set ears, micrognathia (small jaw), congenital heart disease, clenched hands, the a prominent occiput. It is a result of trisomy 18. It occurs at a frequency of 1:8000 and often death occurs within 1 year of birth. It is three times more common in girls than boys. Patau’s syndrome is due to trisomy 13, and it is characterised by severe mental retardation, microphthalmia, microcephaly, cleft lip/palate, coloboma eye, abnormal forebrain structures, polydactyly, and congenital heart disease. The rate of occurrence is 1:6000. Metafemale – trisomy X Turner’s syndrome: Low hairline, broad chest, short stature, retrognathism and webbed neck are features of Turner’s syndrome. In 80% cases the origin of the aneuploidy is from paternal X chromosome; hence the single X chromosome present in a subject with Turner’s is of maternal origin. The incidence of Turner’s syndrome is approximately 1 in 2000 live-born female infants. Random inactivation (see

below) does not occur in cells with a single X chromosome. In general, girls with Turner show a disharmonic IQ profile. The full scale IQ is either comparable to general population or lower by a mean of 10 points (nearly one standard deviation) mostly due to reduced performance IQ (at least 20 points or 1 standard deviation lower) though verbal IQ is preserved. Specific subtests assessing visuospatial processing such as ‘Block Design’ and ‘Object Parental origin of meiotic error leading to aneuploidy. Aneuploidy Paternal % Maternal % Patau 13 85 Edward’s 18 90 Down’s 21 95 Turner’s 45X 20 Klinefelter’s 47 XXY 55

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