

## Unit IV – Problem 9 – Genetics: Cytogenetic Abnormalities Related to Abortion

| Cytogenetic Abnormalities (70% of spontaneous losses, 0.7% of newborns)   |   |  |  |   |
|---|---|--|--|---|
| Autosomal Trisomies   | Monosomy X  | Triploidy (three copies of each chromosome)  | Tetraploidy (four copies of each chromosome)   | Unbalanced Structural Chromosomal Abnormalities   |
| <ol style="list-style-type: none"> <li>1. Detected in 50% of abnormal fetuses.</li> <li>2. Causing loss of pregnancy in 1<sup>st</sup> trimester.</li> <li>3. <b>Represented by:</b> trisomies 16, 15 &amp; 22.</li> </ol>  | <ol style="list-style-type: none"> <li>1. Detected in 20% of abnormal fetuses.</li> <li>2. Causing loss of pregnancy in the 1<sup>st</sup> trimester.</li> <li>3. <b>Represented by:</b> Turner syndrome.</li> </ol>  | <ol style="list-style-type: none"> <li>1. Detected in 15% of abnormal fetuses.</li> <li>2. Causing loss of pregnancy in 1<sup>st</sup> trimester.</li> <li>3. <b>Due to:</b> dispermy, diploid sperm or diploid ovum.</li> <li>4. <b>Causes hydatidiform mole:</b> overgrowth of placenta more than embryonic tissues. It can be:                             <ul style="list-style-type: none"> <li>• <u>Partial:</u> there are fetal parts; trophoblastic hyperplasia; no association with malignancy; triploidy.</li> <li>• <u>Complete:</u> there is trophoblastic hyperplasia and association with malignancy; but no fetal parts and there is diploidy.</li> </ul> </li> </ol> | <ol style="list-style-type: none"> <li>1. Detected in 5% of abnormal fetuses.</li> <li>2. Causing loss of pregnancy in 1<sup>st</sup> trimester.</li> <li>3. <b>Due to:</b> failure of post-conceptual cell cleavage (it will stay as 4n).</li> <li>4. <b>Examples:</b> XXXX-XXYY</li> </ol> | <ol style="list-style-type: none"> <li>1. Detected in 2 % of abnormal fetuses.</li> </ol>   |
| Recurrent Abortion (1-2% of couples experience ≥ 3 consecutive spontaneous pregnancy losses)  |   |  |  |   |
| Luteal Phase Deficiency (LPD)   | Anatomical Defects in The Mother  | Parental Chromosomal Abnormalities   | Underlying Maternal Disease  | Genetic Factors   |
| <ol style="list-style-type: none"> <li>1. A heterogenous disease with a multifactorial causes (can be from hypothalamus/pituitary; ovaries or endometrium).</li> <li>2. Luteal phase &lt; 12 days with normal progesterone level.</li> <li>3. Normal luteal phase with abnormal progesterone levels.</li> </ol> | <ol style="list-style-type: none"> <li>1. Uterine anomalies being the most common:                             <ul style="list-style-type: none"> <li>• Didelphus.</li> <li>• Arcuate uterus.</li> <li>• Uterus bicornis.</li> <li>• Uterus bicornis unicollis.</li> <li>• Cervical atresia.</li> <li>• Vaginal atresia.</li> </ul> </li> </ol> | <ol style="list-style-type: none"> <li>1. Reciprocal or robertsonian balanced rearrangement with recurrence risk of 21:21 robertsonian translocation of 100%.</li> <li>2. Chromosomal inversion (9 &amp; 2): very rare.</li> <li>3. Sex chromosome mosaicism: rare.</li> </ol>   | <ol style="list-style-type: none"> <li>1. Hypertension</li> <li>2. Diabetes</li> <li>3. SLE</li> </ol> And others...   | <ol style="list-style-type: none"> <li>1. X-linked dominant disorders.</li> <li>2. Skewed X-chromosome inactivation (more than 90% of the same parental chromosome).</li> <li>3. Autoimmune disorders (ex. Antiphospholipid antibodies which lead to a higher incidence of thrombotic events).</li> <li>4. Vascular deficiency (of factors V &amp; XII).</li> <li>5. Hyperhomocysteinemia.</li> </ol> |
| Other Causes of Pregnancy Loss  |   |  |  |   |
| Exogenous Factors   | 2 <sup>nd</sup> & 3 <sup>rd</sup> Trimesters Fetal Loss   | Abnormal Uterine Environment   | Multiple Pregnancies   |   |
| <ol style="list-style-type: none"> <li>1. Teratogens (drugs..etc).</li> <li>2. Maternal infections (measles, mumps, rubella, syphilis)</li> </ol>   | Hypertension, renal disease... etc.   | <ol style="list-style-type: none"> <li>1. Oligohydramnios (less amniotic fluid).</li> <li>2. Polyhydramnios (excessive amniotic fluid).</li> </ol>   | -  |   |

