Unit 2 Key Area 4

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| **Antenatal (prenatal) screening** | identifies the risk of a disorder so that further tests and a prenatal diagnosis can be offered. |
| **Examples of Antenatal screening include:**  | Untrasound imaging, biochemical testing, diagnostic testing, amnioscentesis, chorionic villus sampling and rhesus antibody testing. |
| **Ultrasound Imaging is carried out at 8-14 wks to produce** | a dating scan – used to determine the stage of pregnancy and calculate a due date |
| **Ultrasound imaging is carried out at 18-20 wks to produce**  | an Anomaly Scan, which shows up the any serious physical abnormalities in the foetus |
| **Biochemical tests** | monitor physiological changes that occur during pregnancy eg. Concentrations of human chorionic gonadotrophin |
| **Diagnostic testing** | A definitive test that produces definite results about whether or not a person is suffering from a specific condition  |
| **Amnioscentesis and Chorionic Villus Sampling** | Used to prepare a person’s karyotype which shows their complete chromosome complement |
| **Amnioscentesis involves obtaining**  | amniotic fluid containing foetal cells. The cells are then used to create a karyotype, allowing detection of abnormalities |
| **Chorionic villus sampling (CVS) involves obtaining** | a sample of placental cells. The cells are then used to create a karyotype, allowing detection of chromosome abnormalities |
| **Amnioscentesis is carried out**  | Between 14-16 wks |
| **Chorionic villus sampling (CVS) is carried out** | At 8wks |
| **Disadvantage of chorionic villus sampling** | It causes higher incidence of miscarriage than amniocentesis |
| **Pregnant women who are rhesus negative** | will have immune system problems if her baby is Rhesus positive as the fetus red blood cells are seen as “foreign”  |
| **All newborn babies are screened for PKU (phenylketonuria)** | By having their blood tested of presence of excess phenylalanine. |
| **Pedigree charts can be used to** | analyse patterns of inheritance in genetic screening and counselling |
| **An example of autosomal recessive inheritance is** | Cystic Fibrosis |
| **Autosomal recessive inheritance** | Trait is expressed relatively rarely, skips generations and sufferers are homozygous recessive. |
| **Autosomal dominant inheritance** | Trait is expressed every generation, each sufferer has an affected parent and sufferers are homozygous dominant or heterozygous |
| **An example of autosomal dominant inheritance** | Huntington’s disease |
| **An example of autosomal incomplete dominance** | Sickle cell disease |
| **In sex-linked recessive traits** | More males are affected than females, none of the sons of an affected male show the trait and all sufferers are homozygous recessive. |
| **An example of a sex linked recessive trait** | Haemophilia |