




Consideration in Development


- Gene theory – Sickle Cell
- Environmental theory – AN / BN
- Interactionist theory - Phenylketonuria (PKU - mutation on chromosome 12) – Inability break down the amino acid phenylalanine (found in milk and meat), therefore builds up in the blood. Symptoms:
 - Convulsions
 - Hyperactivity
 - Severe mental retardation.
- PKU children appear normal at birth, delay observed post 6months, severe by age 4. If detected at birth, PKU can be controlled by a diet.



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Prenatal Developmental Risks

- The foetus is not immune from the outside world. In fact, some things can be damaging to the unborn child – Teratogens
- Environmental or genetic agents that cause birth defects. For example:
 - **Environmental teratogens**
 - Poor nutrition
 - Stress
 - Pollutants
 - Cigarette smoking
 - Alcohol, and drug abuse
 - **Genetic teratogens:**
 - Related to the heredity of the unborn child:
 - Maternal disease (HIV)



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Prenatal Developmental Risks

- Many factors come into play when considering the health of an unborn child. It is not only important how a mother takes care of her physical self, but her emotional self as well.
- Furthermore, the environment in which a pregnant women lives can greatly affect the health of her unborn baby, e.g.
 - Women who are abused by their partner are more likely to deliver LBW babies.
 - The effect of physical and/or emotional abuse during pregnancy is comparable to the rate of high blood pressure during pregnancy.

Therefore expectant mothers need to take care to minimise the risk of exposure to teratogens.

Parents can also maximize babies development!

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Physical Risk Factors

- Age less than 18 or over 40 – Older women have higher rates of delivery complications, Down's Syndrome.
- Family history of inherited disease
- Crowded living conditions
- Malnutrition – Premature, LBW, delayed development, intellectual deficits, stillbirth
- History of infant over 10 lbs
- History of dysfunctional labour
- Less than 12 months from last delivery to due date
- History of LBW infant (less than 5 lbs.)
- Repeated spontaneous abortions (3 or more)
- History of premature baby (less than 37 weeks)
- Pre-eclampsia (history of)
- Hepatitis (within last two years)
- Previous post-partum haemorrhage
- Diabetes – Foetal death, respiratory difficulties, metabolic disturbances

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Health Behaviour Risk Factors 1

- Smoking greater than one pack per day – low birth weight, delayed development, conduct disorder
- Poor nutrition
- History of previous drug use
- Surviving birth-damaged infant
- Overweight (more than 50 lbs over ideal weight at time of conception)
- Neonatal death (stillborn or within first 28 days)
- Uterine anomaly
- Serious emotional problems
- Previous caesarean section, low horizontal incision
- Medical History Risk Factors
- Rubella (German measles)- 1st trimester – Deafness, cataracts, heart defects, developmental delay
- Hypertension – Miscarriage
- Past uterine surgery (if uterus incised)
- Heart disease
- Chronic pulmonary disease
- Diabetes
- Serious endocrine problems
- Serious psychiatric problems
- Serious neurological problems

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Use and abuse of the following:

- Analgesics, Antithetic, Barbiturates, Antidepressants – Respiratory depression
- Aspirin (large quantities) – Bleeding, spontaneous abortion, respiratory depression
- Alcohol – Developmental delay, heart problems
- Antihistamines – Malformation, foetal death
- Hallucinogens
- Narcotics (heroin, morphine) – Respiratory depression, death, delayed development, convulsions, withdrawal symptoms
- Psychotropic herbs
- Sedatives
- Hypnotics
- Quinine – Deafness
- Tobacco – Spontaneous abortion, prematurity, LBW, convulsions, high heart rate, sudden infant death (SIDS), cleft-lip palate, reduced head circumference
- Thalidomide – Deformed limbs, hearing defects, death

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Effects of drug and alcohol consumption on the foetus 1

- There is evidence to suggest that a substantial amount of alcohol or cocaine can result in a baby being born with Foetal Alcohol Effects (FAE). The most severest of these cases result in Foetal Alcohol Syndrome (FAS).
- Some of the most common and more visibly recognizable effects of FAS:
 - Enlarged forehead
 - A smaller space between the nose and top-lip
 - Droopy-like eyes
 - A smaller head

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Effects of drug and alcohol consumption on the foetus 2

- Some of the most severe birth defects include:
 - Defective Joints
 - Major Heart defects
 - Mental Retardation
 - Hearing defects
- Some of the less severe effects are:
 - An Uncontrollable Temper
 - Hyperactivity
 - Mentally challenged or slower
 - Shorter attention span

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Effects of drug and alcohol consumption on the foetus 3

- About 16% of pregnant women drink enough alcohol to be at risk for bearing children with some sort of negative effect.
- 800 babies/year born with FAS (UK)
- The 1st trimester of pregnancy is the most crucial period - consuming even the smallest amount of drugs or alcohol whilst pregnant raises the risk of miscarriage before the 3rd month.
- Most often, when an infant is born addicted to any substance it is because the mother is not aware that she is pregnant until after most of the damage has been done.

Risk During Prenatal Care 1

- Physical and Health Risk Factors:
 - Diabetes (insulin dependent)
 - Failure to gain weight
 - Heart disease
 - Hepatitis, Herpes, Measles, Mumps, flu, chicken pox, scarlet fever – Death, blindness, deafness, developmental delay, miscarriage
 - Hypertension
 - Anaemia – Stillbirth, death, brain damage
 - Multiple pregnancy
 - Pre-eclampsia
 - Premature rupture of membranes (less than 36 wks & verified dates)
 - Renal disease
 - Rubella after first trimester
 - Serious bacterial or viral infection at term
 - Serious endocrine disorders
 - Sickle cell anaemia
 - Threatened premature labour less than 37 weeks
 - Thromboembolism
 - Weight gain of over 50 lbs

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Risk During Prenatal Care 2

- Physiological Risk Factors:
 - Emotional Stress – High rates of Down's Syndrome, pregnancy complications
 - Failure to follow directions
 - Lack of support
- Environmental Risk Factors:
 - Lead – Anaemia, miscarriage, haemorrhage
 - Radiation – Cancer, leukaemia, cataracts, miscarriage, stillbirth, microcephaly
 - Mercury – Cerebral palsy

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PRENATAL DIAGNOSIS / SCREENING / GENETIC COUNSELLING 1

- ULTRA SOUND (SONOGRAPHY)
 - Ultrasound uses high frequency sound waves directed into the abdomen. The echo from the sound is produced into pictures on a monitor.
 - This type of testing can be used to find foetal weight, length, position, number of foetuses, and various physical features or abnormalities.

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PRENATAL DIAGNOSIS / SCREENING / GENETIC COUNSELLING 2

- AMNIOCENTESIS
 - This is a medical procedure in which a sample of the amniotic fluid is withdrawn with a syringe.
 - The fluid is tested for chromosomal abnormalities, as well as metabolic disorders.
 - It is done between the 12th and 16th week of pregnancy.

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PRENATAL DIAGNOSIS / SCREENING / GENETIC COUNSELLING 3

- CHORIONIC VILLUS SAMPLING (CVS)
 - In this procedure a sample of the placenta is taken between the 8th and 11th week. The results can show chromosomal abnormalities.
- MATERNAL BLOOD TEST (aka alpha-fetoprotein).
 - The blood test can be done in the 14th to 20th week.
 - Neural tube defects can be detected with this type of testing.

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Genetic Disorders in Prenatal Development

- Genetic Codes
 - The information that tells our bodies how to develop is in the form of genetic codes.
 - These codes also carry the disorders that our bodies may or may not develop through life...**However**, this does not mean that we will automatically be affected by these disorders within our lives.
 - A person that does not show the symptoms of the disorder is usually known as a carrier, i.e. a carrier has the genotype to have the disease, but is lacking the dominant gene to infect him or her to the point that symptoms show.
 - In order infect their off-spring with the disorder, carriers must conceive children with another carrier with the full fledged disorder.
 - Infants with active genetic disorders account for approx. 25% of the infant death rate each year.
 - Statistics such as these lead many want-to-be-parents to genetic counselling, which helps determine their risk for having a child with a genetic disorder.
- Screening
 - Abnormalities in the genes can be determined using prenatal tests - Amniocentesis

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Anencephaly

- Anencephaly is a very devastating neural tube disorder, which causes brain and skull malformation.
- Most children die at birth.
- Ultrasound and amniocentesis are used in diagnosis.
- Occurs in the third or fourth week of prenatal development.
- Involves the incomplete development of the brain, spinal cord, and the coverings that protect them due to the neural tube not closing at the cephalic (head) end.
- Babies born without a forebrain and a cerebrum, which often leaves the neonate blind, deaf, unconscious, and unable to feel pain.
- There is no known treatment for it leading to the newborn's death within a few hours or days after birth.
- One possible prevention of the disorder is the intake of dietary folic acid by the mother. It is believed to drop the rate of anencephaly from 1 in 1,000 to 1 in 10,000.



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Anencephaly



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Down Syndrome (Trisomy 21)

- The most common form – Nondisjunctive / trisomy 21.
- Affects 95% of the DS population and occurs because of a faulty cell division, resulting in three 21st chromosome, rather than the normal pair.
- DS is a genetic disorder but not necessarily an inherited disorder.
- Down's includes LD and physical deformities to varying degrees.
- Some of the physical differences are a round face, protruding tongue, short limbs, and motor deficiency.
- High quality of live over past two decades
- Maternal blood tests, CVS, and amniocentesis can be used to detect Downs.



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Tay-Sachs disease

- Hereditary disorder of the NS that causes severe brain damage, enlargement of the head, convulsions, blindness, deafness, lack of energy, and eventually death.
- Physically evident a 6-months, beginning with a reddish dot on the retina of the eye.
- Tay-Sachs disease occurs in children who have too little of the enzyme **hexosaminidase**. This controls the amount of ganglioside that accumulates in the nerve cells. Ganglioside is a lipid produced by normal cell reproduction. Nerve cells that store too much of the lipid become swollen and die. Brain damage can result from a large number of the damaged or dead cells.
- Recessive - Both parents must be carriers of the trait for the child to inherit the chance of being infected fully by Tay-Sachs disease.
- Thus, $\frac{1}{4}$ born by parents that are carriers will have full Tay-Sachs disease, and $\frac{1}{2}$ of the children that are born will be carriers of the disease.



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PKU (Phenylketonuria)

- Recessive genetic disease that, if not detected and treated within the first few weeks of life, leads to developmental delay and other neurological problems.
- The occurrence of children with PKU in UK is about 1 in every 14,000 live births, while the incidence of being a carrier is 1 in 50.
- Individuals must adhere to a very strict diet beginning at birth in order to live a normal and healthy life.
- Diet is **low in protein** (including breast milk) – Protein v. high amounts of phenylalanine – not able to process.
- Normal diet may be resumed approx. 6 y.o. as believed that once the brain was fully developed no more damage could be done by the disease.
 - HOWEVER...being taken off the diet at any time can result in learning disabilities, behaviour problems, and a drop in IQ.
- Women with PKU who are planning on having children must monitor the level of phenylalanine intake in their diet because a high level of phenylalanine in the blood stream may harm the unborn child.

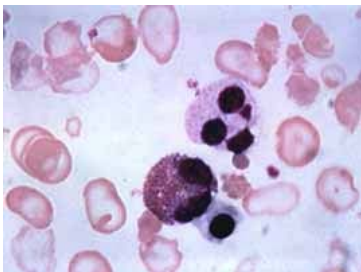
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Thalassemia

- Inherited red blood cell disease
- Primarily due to the inability of the red blood cells to produce haemoglobin which carries oxygen to the body.
 - Without haemoglobin, the body's organs do not get the oxygen they need to sustain normal life.
 - The red blood cells become abnormally thin and produce pale skin, tiredness, irritability, and loss of appetite.
 - The heart becomes enlarged along with the liver and spleen.
 - Also weakens bones (particularly facial)
 - Symptoms of the disease are present at birth or generally appear within 6 months.
- Most people with thalassemia appear jaundiced.
- Thalassemia cannot be cured, but can be treated by blood transfusion and antibiotics. This relieves the symptoms but causes an iron excess in some organs. This iron excess often leads to diabetes and heart failure.

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Thalassemia



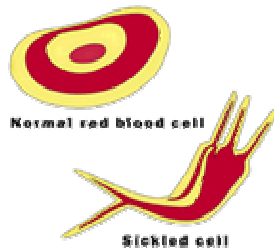
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Sickle Cell Disease and Anaemia

- Inherited genetic abnormality in red blood cells.
 - This is when a person's infected Red Blood Cells (RBC) take the shape of a crescent/sickle shape (instead of disk-shaped).
 - An abnormal RBCs causes problems with that person's ability to use oxygen.
 - Haemoglobin, which carries oxygen throughout the body, is not properly distributed through-out the red blood cells.
- The infected cells have a short survival time and make the infected person anaemic, and therefore can be a very deadly disease.
- The disease is also characterised by acute extremity pain.
- Sickle Cell is predominantly culture specific.
- Testing for Sickle Cell includes amniocentesis and CVS.

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Sickle Cell Disease and Anaemia



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Brain Development and Intelligence

- Evidence that suggests one's intellectual abilities can be positively influenced during the foetal and infant life stages.

IQ AND BREAST FEEDING



- A review of 20 research publications highlight that IQ is a potential benefit to breast-feeding:
 - Breast-fed babies IQ's are 3-5 points higher than formula fed babies.
 - The longer the baby is breast-fed, the greater the benefits to IQ.
 - Higher IQ's attributed to docosahexaenoic acid (DHA) and arachidonic acid (AA) contained in breast milk - not found in feeding formulas.
 - AA is a long chain of fatty acids that support brain development.

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Brain Development and Intelligence

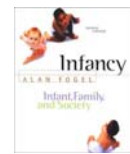
■ Further suggested interventions:

- Regular exercise whilst pregnant (check with G.P.) eases labour.
- No drug use
- Maintain a well nutrition diet
- Reading to unborn child (see Dr. Seuss) (rhythm)
- Playing music to your foetus and child (rhythm)



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