Counselling Station: Down’s Syndrome

- Commonest chromosomal abnormality: 1/650 live births
- Extra chromosome 21- happens prior to fertilization
- Commoner in older mothers although precise mechanism not known: 20 1/2000, 30 1/900, 40 1/100, 47 1/10
- 8% Sometimes not all cells have extra chromosome: mosaicism, cognitive development normal
- 4% caused by translocation carried by mother/father
- DS not preventable, but tests can be done in pregnancy; screening identifies high risk and diagnostic tests confirm via karyotyping

- DS: characteristic facies (upslanting eyes, wide palpebral fissure, prominent tongue), hypotonia, flattened occiput, single palmar crease, curved fifth finger, sandal gap, brushfield spots on iris
- Assoc with developmental delay but IQ usually>80
- Early feeding difficulties
- Increased incidence congenital abnormalities part cardiac & duodenal atresia
- Small stature, hearing problems, visual impairment, learning difficulties

- Many people with DS attend normal school, live fulfilling lives, employment and relationships
- Life expectancy is reduced particularly if there are associated anomalies, but over 50% DS pts live past 50.

- Parental support groups
- Introduce to family of DS pt
- Counselling
- Professional & self-help groups
- Written information

Note
These notes were written by Sarah Mehrtens as a medical student and submitted in 2009. They are presented in good faith and every effort has been taken to ensure their accuracy. Nevertheless, medical practice changes over time and it is always important to check the information with your clinical teachers and with other reliable sources. Disclaimer: no responsibility can be taken by either the author or publisher for any loss, damage or injury occasioned to any person acting or refraining from action as a result of this information. Please inform us of any ambiguities, inaccuracies or errors by emailing bob@askdoctorclarke.com