

Down Syndrome (Trisomy 21)

Condition Description: **Down Syndrome (DS)** is caused by an extra copy of chromosome 21. This is most commonly an independent extra chromosome, but rarely an extra chromosome 21 can be attached (translocated) to another chromosome, involve only a portion of the extra chromosome (partial trisomy 21), or the extra copy is present in only some cells (mosaic trisomy 21). DS is common and the incidence increases in older women¹. Chromosome analysis is required to confirm the diagnosis.

Characteristic features:

1. Typical facial features
2. Post-natal growth delay (use Down syndrome growth charts²)
3. Developmental delay/mental retardation
4. Congenital heart disease (40%)
5. Hearing loss (60-90%. Conductive, sensorineural and combined)
6. Vision abnormalities (Refractive errors very common)
7. Thyroid disease (usually hypothyroid. Lifetime prevalence ~40%)

Other associated findings: Susceptibility to infection (small percentage have IgG subclass deficiency), seizures (5-10%), increased risk for leukemia (10-15 times higher than general population. Routine monitoring not recommended), cataracts, autism, early onset Alzheimer's disease (70-80%).

Differential diagnosis: [Zellweger syndrome](#) (unlikely to be confused although facial features are similar)

Action required: **Chromosome analysis to confirm diagnosis. Echocardiogram in nursery. Consider referral for clinical genetics evaluation, contact [Down syndrome Clinic at Primary Children's Medical Center](#), or general [genetics clinics](#).**
Follow management recommendations.¹⁻⁴

References & Resources:

1. [Facts about Down syndrome. NIH Eunice Kennedy Shriver National Institute of Child Health and Human Development.](http://www.nichd.nih.gov/publications/pubs/downsyndrome.cfm) (http://www.nichd.nih.gov/publications/pubs/downsyndrome.cfm)
2. [AAP Committee on Genetics. Health Supervision for Children with Down syndrome. Pediatrics 107:442-449, 2001](http://aappolicy.aappublications.org/cgi/reprint/pediatrics;128/2/393.pdf) (http://aappolicy.aappublications.org/cgi/reprint/pediatrics;128/2/393.pdf)
3. [Healthcare Guidelines for Individuals with Down Syndrome](http://www.ds-health.com/health99.htm) (http://www.ds-health.com/health99.htm)
4. [Health Care Management of Adults with Down Syndrome. Am Fam Physician 64:1031-1039, 2001](http://www.aafp.org/afp/2001/0915/p1031.html) (http://www.aafp.org/afp/2001/0915/p1031.html)
5. [Genetic Home Reference – Down syndrome](http://ghr.nlm.nih.gov/search?query=Down+syndrome&Search=Search) (http://ghr.nlm.nih.gov/search?query=Down+syndrome&Search=Search)

Patient Resources:

- [National Down Syndrome Society](http://www.ndss.org/) (http://www.ndss.org/)
- [National Association for Down Syndrome](http://www.nads.org/) (http://www.nads.org/)
- [National Down Syndrome Congress](http://www.ndscenter.org/) (http://www.ndscenter.org/)
- [Utah Down Syndrome Foundation](http://www.udsf.org/home0.aspx) (http://www.udsf.org/home0.aspx)
- [What is Down Syndrome?](http://learn.genetics.utah.edu/content/disorders/whataregd/down/) (http://learn.genetics.utah.edu/content/disorders/whataregd/down/)