Down Syndrome

Danielle Keita-Taguchi,
Alex Young
And Lily Clelo-Padua
DOWN SYNDROME
-Trisomy 21

Down syndrome occurs when an individual has three, rather than two, copies of the 21st chromosome.

One in every 733 babies is born with Down syndrome. (Likelihood can increase with age)

The Genetic disorder is named after John Langdon Down
What is Trisomy?

n. A congenital disorder, caused by the presence of an extra 21st chromosome, in which the affected person has mild to moderate mental retardation, short stature, and a flattened facial profile. Also called trisomy 21.
What does a Karyotype of Trisomy 21 look like?

47, XY, +21
Types of Down Syndrome

- Trisomy 21: Child with trisomy 21 has three copies of chromosome 21 — instead of the usual two copies — in all of his or her cells. Caused by abnormal cell division during the development of sperm cell or egg cell.

- Mosaic Down syndrome: rare form children have some cells with an extra copy of chromosome 21, but not all. Caused by abnormal cell division after fertilization.

- Translocation Down syndrome: can occur when part of chromosome 21 becomes attached onto another chromosome, before or at conception. Children with have the usual two copies of chromosome 21, but they also have additional material from chromosome 21 stuck to the translocated chromosome. Uncommon.
Signs of down syndrome

- Children with Down syndrome have a distinct facial appearance. Though not all children with Down syndrome have the same features, some of the more common features are:
  - Flattened facial features
  - Protruding tongue
  - Small head
  - Upward slanting eyes, unusual for the child's ethnic group
  - Unusually shaped ears
  - Broad, short hands with a single crease in the palm
  - Relatively short fingers
  - Excessive flexibility
Screening for Down Syndrome

* Prenatal screening and diagnosis

Older women should generally have screening with amniocentesis. Amniocentesis (routinely performed at 14-16 weeks' gestation) is the most commonly used and most reliable invasive diagnostic test.

Other invasive diagnostic tests include chorion villi biopsy (CVS) in the first trimester and cordocentesis (collection of fetal blood from the umbilical vein with an ultrasound-guided needle). Fluorescence in situ hybridization (FISH) analysis may be performed to analyze interphase cells (uncultured cells) and metaphase spreads (cultured cells) for speedy results. However, these results should be confirmed with chromosome analysis from cultured fetal cells.

Other screening tests include testing for low maternal serum alpha-fetoprotein (MSAFP), high human chorionic gonadotropin (hCG), and low unconjugated estriol (uE3).
Down syndrome may also be suspected based on prenatal ultrasonography in routine examination or in women at high risk. The prenatal ultrasound markers include the following:

- Nuchal (neck) fold thickening - Identifies 75% of Down syndrome fetuses
- Shortened humerus or femur (leg bones) length - Detect about 31% of cases
- Cystic hygroma (cystic structure in neck region)
- Duodenal atresia or stenosis (double bubble sign)
- Cardiac defects - The most common defects are endocardial cushion defect with atrial and ventricular septal defects.
- Echogenic bowel (the bowel reflects sound waves)
- Renal pyelectasis (dilatation of the pelvis of the kidney)
What should be expected?

- About 40 to 50 percent of babies with Down syndrome have heart defects.
- About 10 percent of babies with Down syndrome are born with intestinal malformations that require surgery.
- There is no cure for Down syndrome.
* Talk to doctor about regular checkups
* For emotional support and learning where to get more information, check out organization
* Start exploring types of therapies that might be tried based on the growth and development of child
* Child's rate of growth and development is typically slower than other children of the same age
* Loose ligaments in children with Down syndrome make it easy for them to dislocate bones. Remember to have X-rays for child
* Pursue treatment to teach independency and self-sufficiency.
* Work with physical therapist and doctor to design an exercise program to help increase muscle strength
* Take gradual steps to teaching child how to eat independently
* Take extra time to teach child to dress himself or herself
* Establish daily routine for bathing and getting ready.
* Speech therapy can help children with DS learn necessary skills.
What will my child look like?

Parent 1
Black Hair BBRR
BB= two dominant alleles for black hair
RR= 2 dominant alleles for not red hair

Parent 2
Blonde hair bbRR
bb= 2 recessive alleles for blonde hair
RR= 2 dominant alleles for not red hair
100% chance child will have black hair.
Result will be child with BLACK HAIR
LINKS USED

http://www1.istockphoto.com/file_thumbview_approve/2702091/2/istockphoto_2702091_cute_nine_month_old_asian_baby_boy.jpg
http://www.ucsfhealth.org/childrens/medical_services/preg/prenatal/conditions/down/signs.html
http://www.ds-health.com/pueschel.htm
http://www.lpch.org/DiseaseHealthInfo/HealthLibrary/genetics/downs.html
http://children.webmd.com/tc/down-syndrome-treatment-overview

http://www.webmd.com/hw-popup/controversial-treatments-for-down-syndrome