Down Syndrome

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1. What Is Down Syndrome?

- Down syndrome is a genetic disorder that causes lifelong mental retardation, developmental delays and other problems. Down syndrome varies in severity, so developmental problems range from moderate to serious. • Down syndrome is the most common genetic cause of severe learning disabilities in children, occurring in one in every 700 to 800 infants.

- if one is at high risk of having a child with Down syndrome or has already had one child with Down syndrome, they may wish to consult a genetic counselor before becoming pregnant. • A genetic counselor estimate their chances of having a child with Down syndrome. He or she can also explain the prenatal tests that will be offered and help figure out the pros and cons of testing.

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This additional genetic material alters the course of development and causes the characteristics associated with Down syndrome. A few of the common physical traits of Down syndrome are low muscle tone, small stature, an upward slant to the eyes, and a single deep crease across the center of the palm – although each person with Down syndrome is a unique individual and may possess these characteristics to different degrees, or not at all.

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2. Common Symptoms

Common Symptoms 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**

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Upward slanting eyes, unusual for the child's ethnic group

Unusually shaped ears.

Children with Down syndrome may also have:

Poor muscle tone

Broad, short hands with a single crease in the palm

Relatively short fingers

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Excessive flexibility.

Infants born with Down syndrome may be of average size, but typically they grow slowly and remain shorter than other children of similar age. Children with Down syndrome also have some degree of mental retardation, most often in the moderate range.

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Down Syndrome Features


Features of Down syndrome
Epicanthic folds
Upslanting palpebral features
Low-set small ears
Flat facial profile
Short neck with excess skin
Furrowed tongue
Sandal-toe deformity
Hypoplastic incurved 5th finger
Single transverse palmar crease
Brushfield spots

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3. Causes

The cause of Down syndrome is one of three types of abnormal cell division involving chromosome 21. All three abnormalities result in extra genetic material from chromosome 21, which is responsible for the characteristic features and developmental problems of Down syndrome. The three genetic variations that can cause Down syndrome include:

- **Trisomy 21.** More than 90 percent of cases of Down syndrome are caused by trisomy 21. A child with trisomy 21 has three copies of chromosome 21 — instead of the usual two copies — in all of his or her cells. This form of Down syndrome is caused by abnormal cell division during the development of the sperm cell or the egg cell.

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- **Mosaic Down syndrome.** In this rare form of Down syndrome, children have some cells with an extra copy of chromosome 21. This mosaic of normal and abnormal cells is caused by abnormal cell division after fertilization.

- **Translocation Down syndrome.** Down syndrome can also occur when part of chromosome 21 becomes attached (translocated) onto another chromosome, before or at conception. Children with translocation Down syndrome have the usual two copies of chromosome 21, but they also have additional material from chromosome 21 stuck to the translocated chromosome. This form of Down syndrome is uncommon.

There are no known behavioral or environmental factors that cause Down syndrome.

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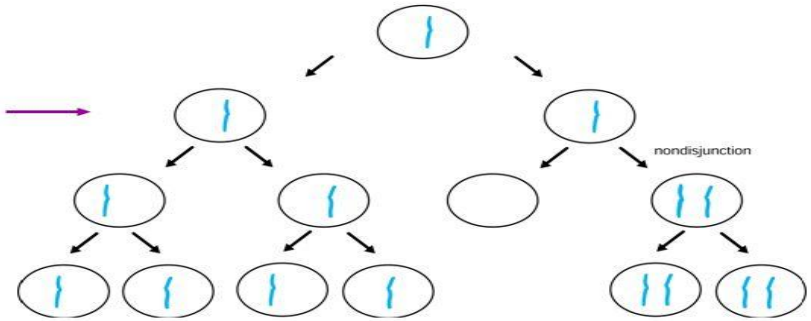
Part of a regular cell showing chromosome 18, 19, 20, and 21 pairs

Part of a Down Syndrome child's cell showing chromosome 21 pair with 1 extra chromosome

Down Syndrome

Down Syndrome: Cytogenetic Causes

- Numerical: ~95%
  - 90% meiotic ndj
  - 5% mosaic 46/47,+21



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4. Risk Factors

Risk of Down Syndrome and Other Chromosome Abnormalities in Live Births by Maternal Age

MATERNAL AGE (AT TERM)	RISK		MATERNAL AGE (AT TERM)	RISK		MATERNAL AGE (AT TERM)	RISK	
	DOWN SYNDROME	TOTAL CHROMOSOME ABNORMALITY		DOWN SYNDROME	TOTAL CHROMOSOME ABNORMALITY		DOWN SYNDROME	TOTAL CHROMOSOME ABNORMALITY
25	1 in 1,250	1 in 476	32	1 in 637	1 in 323	39	1 in 125	1 in 81
26	1 in 1,190	1 in 476	33	1 in 535	1 in 286	40	1 in 94	1 in 63
27	1 in 1,111	1 in 455	34	1 in 441	1 in 224	41	1 in 70	1 in 49
28	1 in 1,031	1 in 435	35	1 in 356	1 in 179	42	1 in 52	1 in 39
29	1 in 935	1 in 417	36	1 in 281	1 in 149	43	1 in 40	1 in 31
30	1 in 840	1 in 385	37	1 in 217	1 in 123	44	1 in 30	1 in 21
31	1 in 741	1 in 385	38	1 in 166	1 in 105	≥45	≥1 in 24	≥1 in 19

Source: Hecht CA and Hook EB. 1996

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Risk Factors Some parents have a greater risk of having a baby with Down syndrome. Risk factors include:

- Advancing maternal age.
- By age 45, the risk is 1 in 35. However, most children with Down syndrome are actually born to women under age 35 because younger women have far more babies.
- A woman's chances of giving birth to a child with Down syndrome increase with age because older eggs have a greater risk of improper chromosome division.By age 35, a woman's risk of conceiving a child with Down syndrome is 1 in 400.

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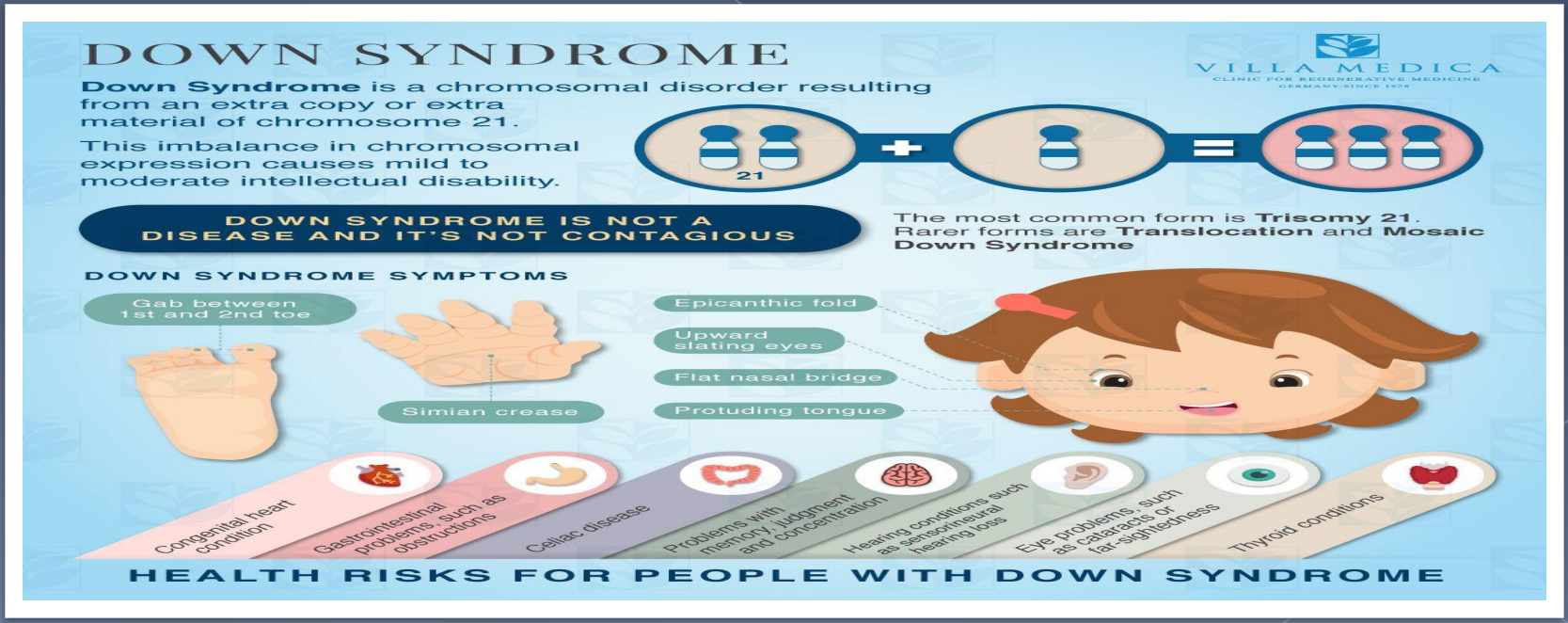
- **Being carriers of the genetic translocation for Down syndrome.**
- Both men and women can pass the genetic translocation for Down syndrome on to their children.



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5. Treatments and Drugs

Early intervention programs

- Ask a doctor about early intervention programs in the area. These specialized programs — in which children with Down syndrome are stimulated at an early age with appropriate sensory, motor and cognitive activities — are available in most states.
- Programs vary from location to location, but they usually involve therapists and special educators whose goal is to help your baby develop motor skills, language, social skills and self-help skills.

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Developing a team

If the child has Down syndrome, you'll likely build a team of specialists that, depending on the child's particular needs, will provide the child's medical care and help him or her develop skills as fully as possible

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- **In addition to your primary care pediatrician, your team may include:**
  - A pediatric cardiologist
  - A pediatric gastroenterologist
  - A physical therapist
  - A speech pathologist
  - An occupational therapist
  - An audiologist
  - A pediatric endocrinologist

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Video: Things People With Down's Syndrome Are Tired of Hearing

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