Birth Defects

Down Syndrome

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KEY POINTS

- Down syndrome is a genetic condition where a person is born with an extra chromosome.
- This can affect how their brain and body develop.
- People diagnosed with Down syndrome can lead healthy lives with supportive care.

What it is

Down syndrome is a condition in which a person has an extra copy of chromosome 21. Chromosomes are small "packages" of genes in the body's cells, which determine how the body forms and functions.

When babies are growing, the extra chromosome changes how their body and brain develop. This can cause both physical and mental challenges.

People with Down syndrome often have developmental challenges, such as being slower to learn to speak than other children.

Distinct physical signs of Down syndrome are usually present at birth and become more apparent as the baby grows. They can include facial features, such as:

- A flattened face, especially the bridge of the nose
- Almond-shaped eyes that slant up
- A tongue that tends to stick out of the mouth

Other physical signs can include:

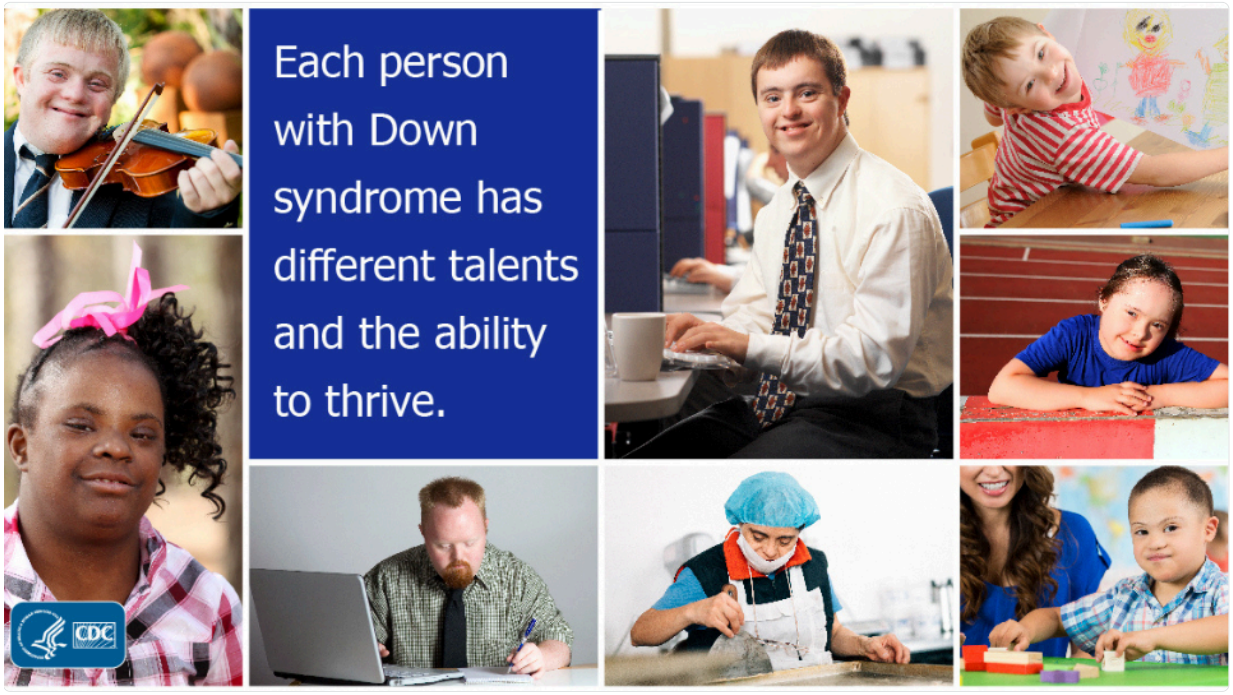
- A short neck
- Small ears, hands, and feet
- A single line across the palm of the hand (palmar crease)
- Small pinky fingers
- Poor muscle tone or loose joints
- Shorter-than-average height

Some people with Down syndrome have other medical problems as well. Common health problems include:

- Congenital heart defects
- Hearing loss
- Obstructive sleep apnea

Down syndrome is the most common chromosomal condition diagnosed in the United States. Each year, about 5,700 babies born in the US have Down syndrome.[1]

KEEP READING:
Living with Down Syndrome



Many people diagnosed with Down syndrome live happy, healthy lives.

Types

There are three types of Down syndrome. The physical features and behaviors are similar for all three types.

Trisomy 21

With Trisomy 21, each cell in the body has three separate copies of chromosome 21. About 95% of people with Down syndrome have Trisomy 21.

Translocation Down syndrome

In this type, an extra part or a whole extra chromosome 21 is present. However, the extra chromosome is attached or "trans-located" to a different chromosome rather than being a separate chromosome 21. This type accounts for about 3% of people with Down syndrome.

Mosaic Down syndrome

Mosaic means mixture or combination. In this type, some cells have three copies of chromosome 21, but other cells have the typical two copies. People with mosaic Down syndrome may have fewer features of the condition. This type accounts for about 2% of people with Down syndrome.

Risk factors

We do not know for sure why Down syndrome occurs or how many different factors play a role. We do know that some things can affect your risk of having a baby with Down syndrome.

One factor is your age when you get pregnant. The risk of having a baby with Down syndrome increases with age. This is especially the case if you are 35 years or older when you get pregnant.[2] [3] [4]

However, the majority of babies with Down syndrome are still born to mothers less than 35 years old. This is because there are many more births among younger women.[5] [6]

Parents with one child with Down syndrome have a higher chance of having another child with Down syndrome regardless of age.[7]

Screening and diagnosis

There are two types of tests available to detect Down syndrome during pregnancy: screening tests and diagnostic tests. A screening test can tell you if your pregnancy has a higher chance of being affected Down syndrome. Screening tests do not provide an absolute diagnosis.

SEE ALSO:
Screening for Birth Defects

Diagnostic tests can typically detect if a baby will have Down syndrome, but they carry more risk. Neither screening nor diagnostic tests can predict the full impact of Down syndrome on a baby.

SEE ALSO:
Diagnosing Birth Defects

Resources

The views of these organizations are their own and do not reflect the official position of CDC.

Down Syndrome Resource Foundation (DSRF): The DSRF supports people living with Down syndrome and their families with individualized and leading-edge educational programs, health services, information resources, and rich social connections so each person can flourish in their own right.

GiGi's Playhouse: GiGi's Playhouse provides free educational, therapeutic-based, and career development programs for individuals with Down syndrome, their families, and the community, through a replicable playhouse model.

Global Down Syndrome Foundation: This foundation is dedicated to significantly improving the lives of people with Down syndrome through research, medical care, education and advocacy.

National Association for Down Syndrome: The National Association for Down Syndrome supports all persons with Down syndrome in achieving their full potential. They seek to help families, educate the public, address social issues and challenges, and facilitate active participation.

National Down Syndrome Society (NDSS): NDSS seeks to increase awareness and acceptance of those with Down syndrome.

SOURCES

CONTENT SOURCE:
National Center on Birth Defects and Developmental Disabilities

REFERENCES

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