Patient Education:

Down Syndrome, Pediatric

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Down syndrome is a genetic disorder that is caused by having an extra chromosome at birth. A chromosome is the cell structure that contains genetic information. A person with Down syndrome is born with part or all of an additional copy of chromosome number 21. Down syndrome may cause certain physical characteristics, affect one's physical and mental development, and cause health problems. However, your child can still lead a very active, successful, and happy life.

CAUSES

Having a full or partial extra copy of chromosome 21 causes Down syndrome. It is not known what causes this extra chromosome to occur.

RISK FACTORS

A person is more likely to have Down syndrome if his or her birth mother was age 35 years or older. The risk of having a child with Down syndrome increases as the mother ages. A person may also be more likely to have Down syndrome if his or her birth mother had previously given birth to a child with Down syndrome.

SYMPTOMS

Physical signs of Down syndrome include:

- Short height (*stature*).
- Small head and ears.
- Short neck.
- Flat nose.
- Large tongue.
- Short, broad hands.
- Large space between the first and second toes.
- Muscles that seem flabby or have a low muscle tone.
- Low-set ears.
- A single line (*crease*) across the palm of the hand (*Palmar crease*).

Children with Down syndrome may also have:

- Developmental delays. They may be slow to crawl or walk.
- Learning disabilities.
- Poor coordination.
- Psychiatric conditions.

DIAGNOSIS

Down syndrome can be diagnosed before and after a child is born. A woman can have prenatal screening tests that check for the likelihood of Down syndrome, but these screening tests do not

diagnose Down syndrome. These can show if there are signs of Down syndrome that need more testing. These screening tests include a blood test and ultrasound. Tests that confirm a diagnosis include:

- Chorionic villus sampling (CVS). This test checks for chromosomal problems during weeks 9–14 of pregnancy.
- Amniocentesis. This test checks for proteins that could indicate birth defects, such as Down syndrome, during weeks 15–20 of pregnancy.

After a child is born, Down syndrome can be diagnosed based on physical appearance. A blood sample may also be taken to check the child's chromosomes. This test can confirm the diagnosis of Down syndrome.

TREATMENT

There are many possible treatments for Down syndrome. Your child's treatment will depend on his or her current symptoms and any other symptoms that develop over time. Treatment almost always requires a team of health care providers and support from other caregivers. Your child's treatment plan may include:

- A heart specialist (*cardiologist*).
- A digestive system specialist (*gastroenterologist*).
- Physical therapy for bone or joint problems.
- Mental health providers or special education teachers to help with any behavioral or learning problems.
- A skin specialist (*dermatologist*).
- A brain specialist (*neurologist*) if your child has neurological changes, such as seizures.
- An eye specialist (*ophthalmologist*) if your child has eye problems, such as cataracts.
- A hormone levels specialist (*endocrinologist*) if your child has thyroid problems.
- A cancer specialist (*oncologist*) if your child develops leukemia.
- A sleep specialist if your child has sleeping problems.

HOME CARE INSTRUCTIONS

- Learn as much as you can about your child's condition.
- Give medicines only as directed by your child's health care provider.
- Work closely with your child's team of health care providers.
- Make sure that you have a good support system at home. This is very important.

SEEK MEDICAL CARE IF:

- Your child has new symptoms.
- You do not have enough support at home.

This information is not intended to replace advice given to you by your health care provider. Make sure you discuss any questions you have with your health care provider.

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