World Down syndrome Day

Down syndrome : A genetic disorder characterized by a variety of physical and mental abnormalities caused by extra genetic material from chromosome 21.

Symptoms

Children and adults with Down syndrome have distinct facial features. Though not all people with Down syndrome have the same features, some of the more common features include:

- Flattened face
- Small head
- Short neck
- Protruding tongue
- Upward slanting eye lids (palpebral fissures)
- Unusually shaped or small ears
- Poor muscle tone
- Broad, short hands with a single crease in the palm
- Relatively short fingers and small hands and feet
- Excessive flexibility
- Tiny white spots on the coloured part (iris) of the eye called Brushfield's spots
- Short height

Risk factors

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

- Advancing maternal age: 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. A woman's risk of conceiving a child with Down syndrome increases after 35 years of age. However, most children with Down syndrome are born to women under age 35 because younger women have far more babies.
- 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.
- Having had one child with Down syndrome. Parents who have one child with Down syndrome and parents who have a translocation themselves are at an increased risk of having another child with Down syndrome. A genetic counsellor can help parents assess the risk of having a second child with Down syndrome.

Complications

- Congenital heart disease
- Upper respiratory infection
- Obstructive sleep apnea
- Gastrointestinal obstruction
- Thyroid dysfunction
- Immune disorders
- Leukemia
- Spinal problems

Diagnosis

Serum TSH is the primary test for diagnosis of thyroid dysfunction.

Once prenatal screening identifies that a pregnant women is at risk for having a fetus with a chromosomal abnormality, cytogenetic analysis can be performed to make a definitive diagnosis. Prenatal samples for cytogenetic testing can be obtained through chorionic villus sampling, usually done at 10 to 12 weeks' gestation, or amniocentesis, done between 15 and 20 weeks' gestation

Studies that have compared rapid trisomy testing, using polymerase chain reaction (PCR) or fluorescence in-situ hybridization (FISH) techniques, with full karyotype analysis have found that rapid testing may fail to detect chromosomal abnormalities in a substantial number of pregnant women.

Cytogenetic analysis can also be performed postnatally if the infant has the characteristic physical appearance of Down syndrome

Monitoring Therapy

Serum TSH is the primary test to evaluate the efficacy of thyroid hormone replacement in patients with primary hypothyroidism.

Perform TSH 4 to 8 weeks after initiating therapy or after a change in dose.

After an adequate replacement dose is determined, measure TSH after 6 months, then at 12 month intervals (more frequently if clinically necessary).

In patients receiving levothyroxine, measure serum TSH within 4 to 8 weeks of initiation of therapy with drugs that decrease the bioavailability of alter the metabolic disposition of levothyroxine.

For women with hypothyroidism who become pregnant, measure serum TSH promptly after conception.

Testing

- Prenatal screening for chromosomal abnormalities in at-risk pregnancies
- Sampling of chorionic villus: Chorionic villus sampling is an accurate method of diagnosing prenatal cytogenetic abnormalities, but accuracy is somewhat less than that of amniocentesis.
- Prenatal screening for chromosomal abnormalities in at-risk pregnancies
- Diagnostic amniocentesis
- Prenatal and postnatal diagnosis of suspected Down syndrome
- Chromosome analysis

Treatment

Medical management of Down syndrome is focused on an organized approach of assessing the patient for heart disease, hearing loss, and ophthalmological disorders, ongoing monitoring for associated disorders such as celiac or thyroid disease, prevention of obesity and periodontal disease, and vigilance in diagnosing and treating other medical conditions found more frequently in patients with Down syndrome, such as diabetes mellitus, leukemia, obstructive sleep apnea, or atlantoaxial subluxation. Providing counseling and resources to the family may help the patient live independently, maintain a job, and interact with others .

Surgical intervention may be necessary in patients with symptomatic atlantoaxial subluxation, congenital heart defects, conductive hearing loss, and obstructive sleep apnea. Surgical procedures for conductive hearing loss include pneumo eustachian tubes, tonsillectomy, and adenoidectomy .

Antibiotic prophylaxis against infective endocarditis for dental procedures should be recommended only for persons with Down syndrome who have congenital heart disease associated with the highest risk of adverse outcome from endocarditis, i.e., complex cyanotic heart disease and postoperative palliative shunts, conduits, or other prostheses.

Reference: Micromedex's Care Notes System Online 2.0