Down Syndrome

Down Syndrome is a chromosomal abnormality. It results in a number of findings including: mental retardation and abnormal physical features. Most cases of Down Syndrome (DS) are derived from the mother's chromosome. About 95% of cases have an extra 21st chromosome and 95% of these are from the mother. It is much more common in older, 35 year old and older, pregnant women. The older the woman is when she gets pregnant, the greater the risk of having a DS baby. There is an incidence of about 1 in 2000 births of DS in mothers less than 20 years of age. Only 20% of DS children are born to mothers under 35 years of age. There are a number of methods of inheritance, and a caregiver or geneticist can discuss these with you. A male DS individual cannot reproduce.

Many people with DS generally live to age 50 or 60 years. This is often decreased because of heart disease and susceptibility to leukemia. As they age, they are prone to developing hearing and visual problems, so regular screening is helpful. Thyroid problems are also common and should be looked for. Almost all individuals with DS show signs of a type of brain illness (*dementia*) beginning in their 30's.

There are many wonderful educational and developmental programs for these children, allowing them to live full and satisfying lives.

CAUSES

Everyone has 23 pairs of chromosomes, 1 from each parent. In DS there are 3 chromosomes on the 21st pair instead of 2. This causes the mental and physical abnormalities to develop.

SYMPTOMS AND SIGNS OF DS

- Mental Retardation.
- Physical Abnormalities:
  - Small head.
  - Heart defects.
  - Small ears.
  - Cataracts.
  - Flat nose.
  - Open mouth with large protruding tongue.
  - Large space between the first and second toes.
  - Short broad hands.
  - Flabby muscles and uncoordinated movements.
  - Fifth finger only has two bones with the little finger curving inward.

DIAGNOSIS

- During pregnancy:
  - Blood tests.
  - Amniocentesis.
  - Chorionic villus sampling.
  - Ultrasound.
  - Percutaneous Umbelical Blood Sampling.
- After delivery or At birth:
  - Physical features mentioned above.
  - Blood tests on the chromosomes.

TREATMENT

- TREATMENT REVOLVES AROUND THE PROBLEMS THAT DEVELOP WITH DS.

- Ear infections.
- Cataracts.
- Thyroid problems.
- Heart defects.
- Leukemia.
- Music therapy seems to help with improving their speech, muscle coordination, learning, social behavior, listening better and being more attentive to people and their surroundings.

PREVENTION

- There is nothing anyone can do to prevent DS.
- If you had a DS baby or are at high risk for having a DS baby, you should consult a doctor specializing in genetics.

HOME CARE INSTRUCTIONS

- DS children need very close and attentive supervision.
- Treat infections of the ears, lungs and intestine quickly because they are prone to complications.
- Play soft and melodic music for them when ever possible.
- Make sure they are examined regularly for thyroid problems, cataracts and heart problems.
- Join support groups with families who have children with DS.

SEEK MEDICAL CARE IF

- The child has a fever 100º F (37.8º C) because of the high risk of ear and lung infections in DS children.
- Your child develops shortness of breath.
- You child has difficulty with vision.
- You notice the child is pale and/or tired.
- The child has behavior problems that you need help with.

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