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Down Syndrome and its Character analysis

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Abstract

Down syndrome one of the common disorder with huge medical condition. DS is associated with phenotype including congenital diseases. DS disorder is a chromosomal condition that mainly happens in case an error is experienced during cell division resulting in an extra chromosome 21. The inexact IQ of a youngster with DS disorder is around 50, equivalent to an 8-9 years of age kid's intellectual ability. In the present review article, we emphasize on the DS, History and physical and congenital development.

Index Terms: Nuchal translucency (NT), Human chorionic gonadotropin (HCG), plasma protein-A (PAPP-A).

Introduction

DS is a chromosomal disorder resulting from the existence of an extra copy choromosome21. This got its name from the doctor who has first described it, John Land Down.

Complete trisomy 21. In this case, an error during the formation of the egg or the sperm result in either one having an extra chromosome. So, after the egg and sperm unite, the resulting cells will also have three copies of chromosome 21. The complete extra copy of chromosome 21 is in all of the person's cells-or a complete trisomy. Almost all Down syndrome cases result from complete trisomy 21. (1,2)

Symptoms of Down Syndrome:

Each person with Down syndrome is having intellectual and developmental issues. That may mild, moderate or severe. Even some are having major problems such as heart defects.

In down syndrome, adults and children are distinct facial features. These are as follows:

*Flattened face

*Small Head

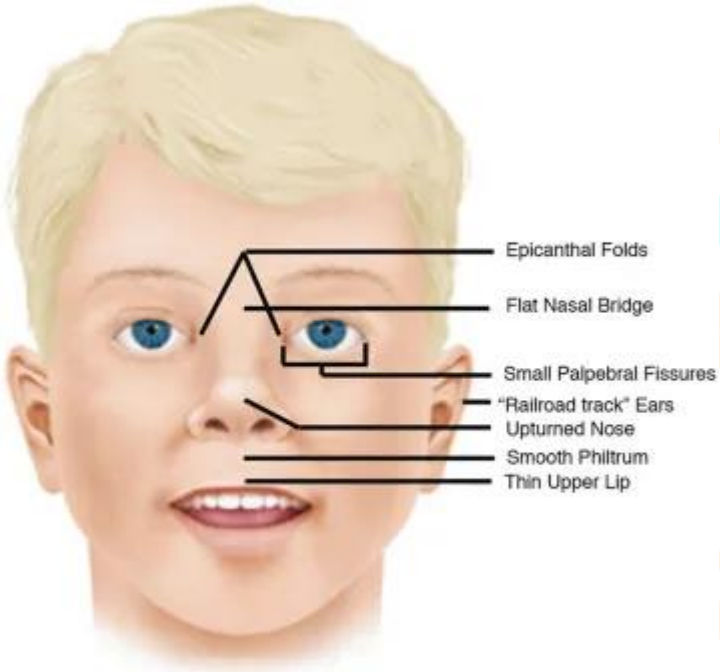
*Short Neck

*Protruding Tongue

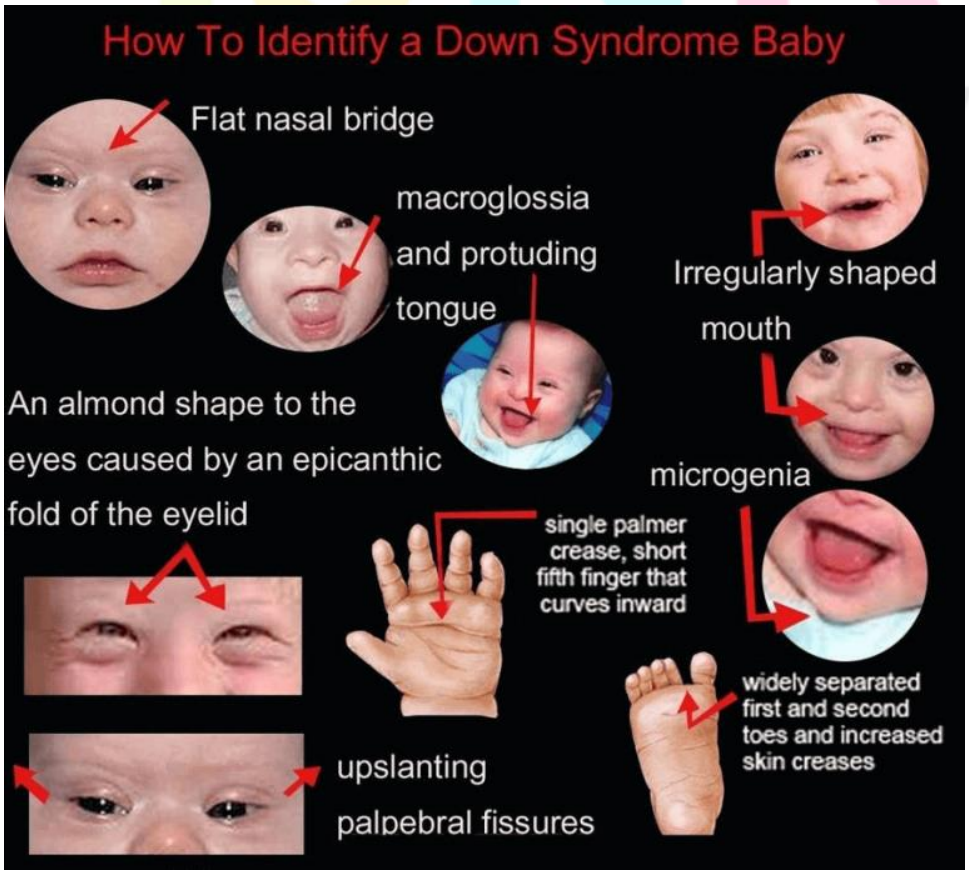
*Upward slanting eye lids (palpebral fissures)

*Unusually shaped or small ears

*Poor muscle tone



Down Syndrome - Pictures, Life Expectancy, Symptoms, Facts, Treatment. 4



Infant with Down Syndrome can be average in size, but mostly they grow very slowly and remain shorter than other children of their age.

Diagnosis of Down Syndrome

SCREENING TESTS DURING PREGNANCY

This test is for down syndrome which considered as a part routine prenatal care. Moreover, Screening test only identify your risk of carrying a baby with the syndrome, which helps you to make decisions for more specific diagnostic test.

It includes:

- The First Trimester combined test
- The Integrated screening tests

`The First trimester Combined Test:`

This process has two steps included,

- Blood Test: This blood test measures the levels of pregnancy-associated plasma protein-A (PAPP-A) and the pregnancy hormones known as human chronic gonadotropin (HCG). Abnormal levels of PAPP-A and HCG may indicate a problem with the baby.
- Nuchal translucency test: Nuchal translucency means, an ultrasound is used to measure specific area on the back of your baby's neck.

`Integrated Screening Test:`

The integrated screening test is done in two parts during the first and second trimester of pregnancy. The results are combined to estimate the risk that your baby has down syndrome.

*First Trimester: part one includes a blood test to measure PAPP-A and an ultrasound to measure NT.

*Second Trimester: The quad screen measure your blood level of four pregnancy-associated substances: alpha fetoprotein,estriol, HCG and inhibin A.

Test name	DR @ SPR = 1%	DR @ SPR = 3%	DR @ SPR = 5%
Double test (using free-β hCG)	46%	63%	71%
Triple test (using free-β hCG)	56%	70%	77%
Quadruple test (using free-β hCG)	66%	79%	84%
Combined test	66%	78%	83%
Serum integrated test	77%	86%	90%
Integrated test	84%	91%	93%

Detection rates for different antenatal Down syndrome screening strategies from the SURUSS. (4)

Preventions of Down syndrome:

There is no way to prevent Down syndrome. If you are at high risk of having a child with Down syndrome or you already have one child with Down syndrome, you may want to consult a genetic counselor before becoming pregnant.

Conclusion

Down Syndrome is being the commonest chromosomal abnormality among live bound infants, it is a number of congenital malformations. It was believed to be responsible for several Down syndrome phenotype including craniofacial abnormality, congenital heart defects, mental retardation and several features.

Since many clinical managements needed in Down syndrome, hence it is important to monitor these patients continuously. Which has been reviewed in the article.

References:

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