



Pathophysiology Concept Map

Assigned Alteration: Down Syndrome AKA Trisomy 21

Why Does it Happen? Risk Factors

Down Syndrome occurs as an error in cell division. The resulting error creates an extra copy of chromosome 21.

The error, disjunction, causes chromosomes to be displaced in cell division.

This usually happens in the formation of the egg or sperm.

What is Wrong?

Pathophysiologic Alteration

Numerous physical alterations. Includes shortened extremities and other facial alterations.

Short hands, marked by a Simian crease (single palmar crease across the hand).

Hyperflexible joints. Low muscle tone.

Dry skin and premature aging.

At risk for congenital heart defects.

What Cues Should the Person Have? Expected Findings

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Trisomy 21

How is it Diagnosed? Laboratory and Diagnostic Tests

Diagnosis of Down Syndrome occurs after a screening suggests the likelihood during pregnancy.

A diagnosis is reached by removing a sample of genetic material from the fetus, and checking for extra material from chromosome 21.

A diagnosis can also be reached through samples of amniotic fluid, placenta, umbilical blood, or maternal blood.

Physical symptoms can be used to diagnose patients after birth.

How is it Managed?

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Those with Down Syndrome suffer from heart problems that can lead to premature death. They are also at an elevated risk for Alzheimer's disease. Those with Down Syndrome will go through a

What Cues Should the Person Have? Expected Findings

Flattened face or facial appearance and flattened posterior skull.
Accelerated aging. This can drastically reduce their life expectancy,
(Brachycephalic head)

Flat nasal bridge. Epicanthal folds (at birth). Upward slanting eyes,
or palpebral fissures.