22q11.2 Deletion Syndrome

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IASSIDD INFORMATION SHEET

22q11.2 Deletion syndrome, as its name suggests, occurs when a small part of chromosome 22 is deleted.

- The deletion is on the long arm (known as the q arm) at a location known as 11.2, near the middle of the chromosome.
- This deletion results in the poor development of several body systems.

Who is affected?

- Occurs between 1 in 4000 to 1 in 6000 live births/people.
- Affects males and females equally.
- Affects all ethnicities and races.
- Is the most common microdeletion syndrome.

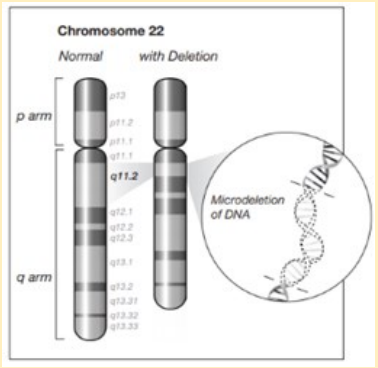
Cause

The exact cause of the small deletion (microdeletion) is not known. It occurs as a random event in the development of the father's sperm or the mother's egg, or early during fetal development at the embryonic phase.

Once it does occur, it may be passed on by one parent to a child, with a 50% chance of inheritance. About 10-15% of those living with 22q11.2 deletion syndrome have inherited the condition from a parent.

Diagnosis

When 22q11.2 deletion syndrome is suspected, it should be confirmed with genetic testing. Corroborating tests include: echocardiogram (cardiac ultrasound), kidney ultrasound, x-rays, and blood tests (CBC, calcium).



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Features and Characteristics

The features and characteristics of 22a111.2 Deletion syndrome are numerous and vary considerably from one person to another, but common features are:

Newborns

- Weak heartbeat
- Low muscular tone
- Long narrow face, small mouth
- Small low-set ears
- Scoliosis (spine curvature)

Early and middle childhood

- Feeding problems
- Speech difficulties
- Short stature
- Frequent infections
- Middle ear infections; hearing loss.
- Vision problems
- Seizures

Childhood and adolescence

- Learning disorders, especially with visual materials; ADHD
- Developmental delays
- Communication and social interaction problems; ASD-like symptoms

Interventions

Specific intervention methods should match symptoms and needs. General approaches are:

1. Treat medical / physical needs
2. Use behavioural approaches
3. Treat mental health
4. Provide adult living support

More specific guidelines are:

Body structure

- Correct / treat body problems
- Reinforce natural body functions
- Use therapeutic equipment

Physical health

- Correct / treat health problems
- Monitor risk areas closely
- Change the environment

Mental health

- Watch for changes
- Treat the symptoms
- Change the environment

Learning

- Assess in an ongoing way
- Provide educational support
- Engage in direct teaching

Other names...

22q11.2 Deletion syndrome is known by several other names, most commonly DiGeorge syndrome or CATCH 22. It is sometimes abbreviated to simply 22q.

Sources and Further Information

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