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Case Report

A Comprehensive Review of Chromosome 4q Deletion Syndrome

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Abstract

Chromosome 4q deletion syndrome is a chromosomal disorder caused by the loss of a segment of the long arm of chromosome 4. It was first described in 1967 and is associated with symptoms in several organ systems. Some children with 4q deletion syndrome have psychiatric symptoms. These include aggression, hearing passive speech (verbal hallucinations), mood swings, and delusions. It should be noted that not all people with 4q deletion syndrome have these symptoms and the symptoms can vary greatly.

Key words: chromosome 4q deletion syndrome; abnormality chromosomes; genetic disorder

Overview of Chromosome 4q Deletion Syndrome

Chromosome 4q deletion syndrome is a chromosomal disorder caused by the loss of a segment of the long arm of chromosome 4. It was first described in 1967 and is associated with symptoms in several organ systems. The patient may have a very prominent forehead (forehead kiss), enlargement of the back of the head, low-set ears, short hands and wide feet, unusually small size with slow or delayed growth, congenital heart defects, and possible be mental retardation [1].

Clinical Signs and Symptoms of Chromosome 4q Deletion Syndrome

Patients with chromosome 4q deletion syndrome may have the following symptoms: an unusual skull shape, a short nose with an unusual bridge, low-set ears that may not be well formed, a cleft in the roof of the mouth, a short sternum, poor growth, or Developmental delay, moderate to severe mental retardation, heart defects, abnormal heart rhythm (arrhythmia), defective urinary and reproductive organs (urinary dysfunction), small size, small hands and feet, unusually wide eyes (hypertelorism), Pointed fifth finger and unusual nail and decreased muscle tone (hypotonia) which is common. Some children with this disorder may have a small jaw (micrognathia), which leads to breathing problems, and there may be unusual brain (corpus callosum) findings. In some patients, slow growth and mental retardation may be present without obvious physical abnormalities, making diagnosis of the disorder difficult [1,2].

Some children with 4q deletion syndrome have psychiatric symptoms. These include aggression, hearing passive speech (verbal hallucinations), mood swings, and delusions. It should be noted that not all people with 4q deletion syndrome have these symptoms and the symptoms can vary greatly [1,2].

Figure 1: Image of the hands and feet of a child with chromosome 4q deletion syndrome.[1]

Etiology of Chromosome 4q Deletion Syndrome

Genetic information, DNA, is embedded in almost every cell in the body and provides the instructions for how each tissue should function. DNA contains genes that are responsible for making proteins that have specific functions in certain tissues of the body. This DNA is tightly packed into structures called chromosomes. Most people are born with 23 pairs (46 total) of chromosomes in every cell of the body. Each of these 46 chromosomes has a long piece called q arm and a short piece called p arm. Chromosome 4q deletion syndrome is caused by missing part of the long arm of chromosome number 4. This chromosomal deletion is not usually inherited, but occurs for the first time in an affected individual. However, it is possible to pass the deletion mutation to children [1,3].

Symptoms can vary greatly, even in members of the same family. The severity and type of symptoms depend on the size and location of the missing chromosomal fragment. Whether it is interstitial (located between other parts of the chromosome) or terminal (at the end of the chromosome) usually gives information about what to expect. Some of the genes that may be involved in this syndrome are listed in table 1. Researchers are investigating other genes that may play a role in chromosome 4q deletion syndrome [1,3].

Sings	Gene Name
Abnormal bone formation, short stature	BMP3
Unusual facial features or skull shape	SEC31A
kidney problems	PKD2
Seizures, decreased muscle tone, delayed motor development (such as sitting, walking)	GRID2, NEUROG2
heart problems	ANK2, HAND2, HPGD, TLL1
Abnormally shaped hands and feet	FGF2

Table 1: Names of genes involved in chromosome 4q deletion syndrome with corresponding clinical symptoms.

Frequency of Chromosome 4q Deletion Syndrome

Chromosome 4q deletion syndrome is a rare disorder that is present at birth and is estimated to occur in 1 in 100,000 people. Chromosome 4q deletion syndrome can sometimes be detected before birth with prenatal ultrasound and chromosome analysis. The disease affects men and women in equal numbers. It is not currently known whether it affects these ethnicities more than other ethnicities. Usually there is no family history of this disease and it occurs randomly [1,4].

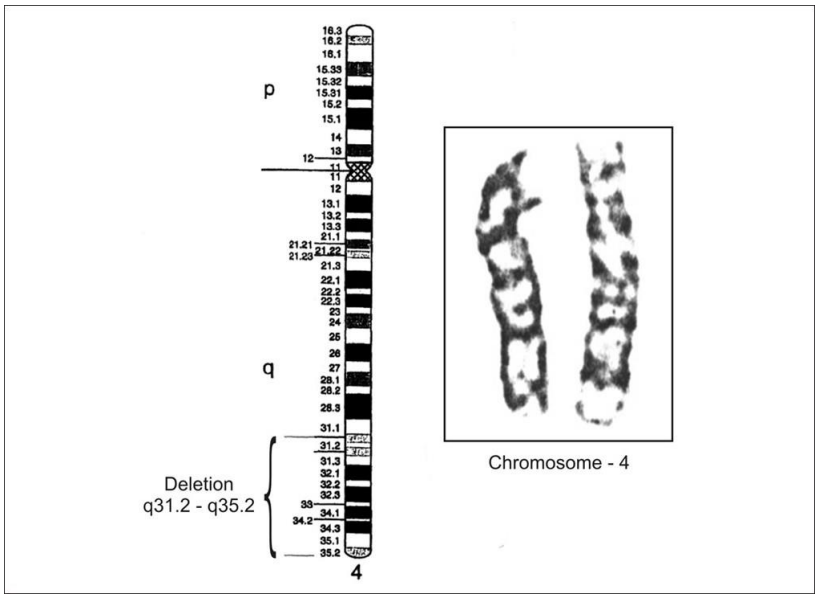

Figure 2: Schematic of Chromosome 4q Deletion.[1]

Disorders Associated with Chromosome 4q Deletion Syndrome

Other chromosomal abnormalities may be similar to chromosome 4q deletion syndrome. Comparison may be useful for differential diagnosis:

Wolf-Hirschhorn syndrome (4p syndrome) is a chromosomal disorder caused by partial deletion of part of the short arm of chromosome 4. The main symptoms include very wide eyes (ocular hypertelorism) with a wide or beaked nose, small head (microcephaly), malformed ears, mental and developmental defects, cardiac (cardiac) defects, and seizures [1,5].

Chromosome 11q monosomy is a rare genetic disorder that affects the long arm of chromosome 11. Symptoms may include eyes that are too far apart (hypertelorism), drooping eyelids, abnormal deviation of the eyes (strabismus), and abnormal eye position. Symptoms may also include a narrow, raised forehead, broad nasal bridge, short upturned nasal tip, carp (fish-like) mouth, receding chin, unusual ears, unusual folds in the palms (candle folds), and mental retardation [1,5].

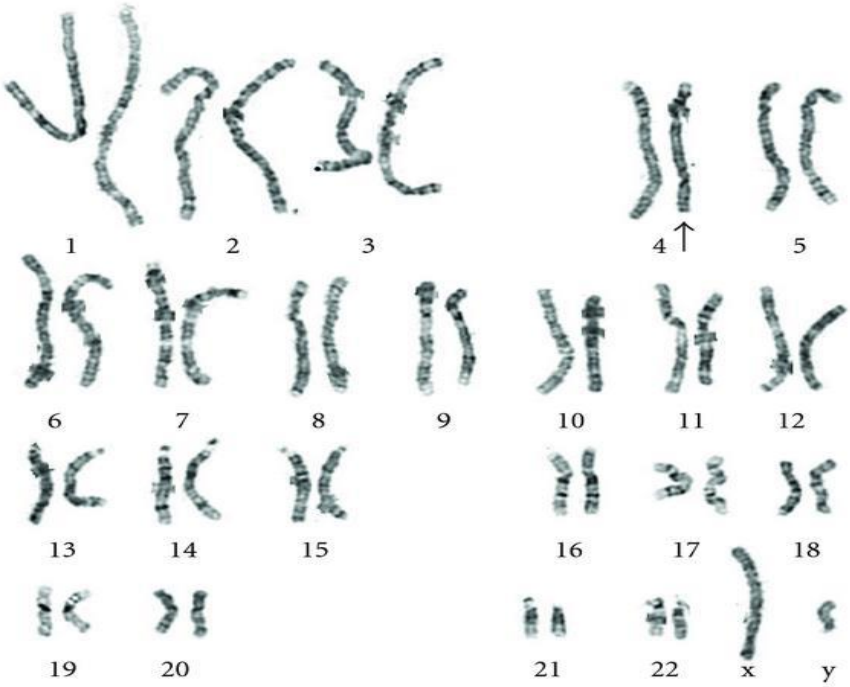

Figure 3: Schematic of Human Karyotype with Deletion of Chromosome 4q.[1]

Cephalopolysyndactyly Grieg syndrome is a rare genetic disorder with symptoms that can include an enlarged head, unusual facial features, and abnormally shaped hands and feet. Some genetic conditions that involve extra chromosomes (trisomy) may also show symptoms similar to deletion of chromosome 4q [1,6].

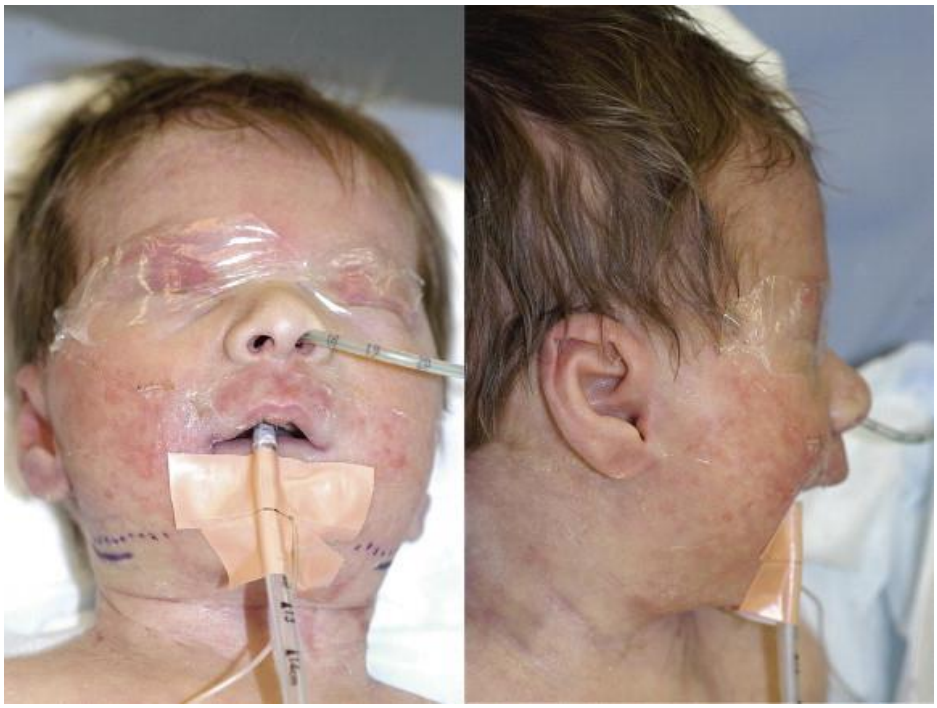

Figure 4: Image of a baby with chromosome 4q deletion syndrome.[1]

Diagnosis of Chromosome 4q Deletion Syndrome

Chromosome 4q deletion syndrome is diagnosed by genetic testing. There are various genetic tests that can be used to find a deletion of chromosome 4q. This includes the following:

1. Karyotype: This test makes a picture of a person's chromosomes. This test can be used to find large deletions.

2. FISH (Fluorescence in situ hybridization): This test is used to find a specific DNA region on a chromosome. A chromosome is attached to a small DNA region called a probe, which has a fluorescent molecule attached to it. This test can be used to find deletions that are too small to be seen on a karyotype. FISH is only useful if the person ordering the test thinks a specific region of 4q has been deleted.

3. Array CGH (Comparative Genomic Hybridization): This test can find deletions that are too small to be seen on a karyotype [1,6].

Treatment Pathways for Chromosome 4q Deletion Syndrome

If needed, surgery is used to treat heart and skeletal problems. Children with delays in walking or other milestones may need treatments such as occupational therapy or physical therapy. Children with small jaws that make breathing difficult may need surgery to lengthen the bones. Medications may be helpful for seizures, reflux, or growth problems. Special education is beneficial for children with intellectual or learning disabilities. Multivitamins and citrus fruits are recommended for all children with 4q deletion syndrome. Coenzyme Q10 can be helpful for people who struggle with slow growth and reflux [1,6].

For patients with chromosome 4q deletion syndrome, special educational and professional services may be helpful. Early intervention services such as physical therapy, occupational therapy, and special education are important. Other treatment is based on individual symptoms [1,6].

Genetic counseling may be beneficial for patients and their families. Genetic counseling involves talking with a specialized health care provider called a genetic counselor who takes the personal and family history of a person with chromosome 4q deletion syndrome. This can be helpful in determining what treatments and resources may be helpful, as well as who in the family should consider genetic testing for chromosome 4q deletion syndrome. Counselors can also explain the chances of a genetic disease running in the family [1,6].

Discussion

Chromosome 4q deletion syndrome is a chromosomal disorder caused by the loss of a segment of the long arm of chromosome 4. It was first described in 1967 and is associated with symptoms in several organ systems. Patients with chromosome 4q deletion syndrome may have the following symptoms: an unusual skull shape, a short nose with an unusual bridge, low-set ears that may not be well formed, a cleft in the roof of the mouth, a short sternum, poor growth, or Developmental delay, moderate to severe mental retardation, heart defects, abnormal heart rhythm (arrhythmia), defective urinary and reproductive organs (urinary dysfunction), small size, small hands and feet, unusually wide eyes (hypertelorism), Pointed fifth finger and unusual nail and decreased muscle tone (hypotonia) which is common. Some children with this disorder may have a small jaw (micrognathia), which leads to breathing problems, and there may be unusual brain (corpus callosum) findings. Children with small jaws that make breathing difficult may need surgery to lengthen the bones. Medications may be helpful for seizures, reflux, or growth problems. Special education is beneficial for children with intellectual or learning disabilities. Genetic counseling may be beneficial for patients and their families. Genetic counseling involves talking with a specialized health care provider called a genetic counselor who takes the personal and family history of a person with chromosome 4q deletion syndrome [1-6].

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