HEMIFACIAL MICROSOMIA

Otherwise known as?

Hemifacial microsomia is part of a broader spectrum of conditions known as craniofacial microsomia, which includes Goldenhar syndrome when additional eye and spinal anomalies are present.

Signs & Symptoms

Hemifacial microsomia (HFM) is a congenital condition where one side of the face is underdeveloped or smaller than the other. This condition primarily affects the craniofacial structures (head and face) and can vary significantly in severity. Below are the signs and symptoms commonly associated with hemifacial microsomia:

1. Facial Asymmetry

- Uneven appearance: One side of the face is smaller or underdeveloped compared to the other, leading to facial asymmetry.
- Jaw and cheekbone differences: The mandible (lower jaw) and zygoma (cheekbone) on the affected side may be smaller or absent.
- Flattened cheek: The cheek on the affected side may appear flatter or recessed.

2. Ear Anomalies

- Microtia: The ear on the affected side may be underdeveloped or not appropriately formed, sometimes referred to as microtia. The ear may be smaller than normal, or missing parts, including the outer ear (auricle).
- Atresia: The ear canal may be absent or underdeveloped, a condition known as aural atresia. This can lead to hearing loss, as the ear canal plays a crucial role in sound conduction.

3. Mouth and Jaw Anomalies

- Cleft lip and/or palate: Some individuals with hemifacial microsomia may also have a cleft lip, cleft palate, or both. This affects the roof of the mouth and/or the upper lip.
- Dental problems: There may be missing teeth, misaligned teeth, or differences in dental development on the affected side.

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4. Eye Anomalies

- Coloboma: A variation in eye formation where a gap or notch occurs in structures like the iris, retina, or optic nerve, can sometimes occur in individuals with hemifacial microsomia.
- Epibulbar dermoids: Benign tumours can form on the surface of the eye in some cases.
- Underdeveloped eyelids: The eyelids on the affected side may be smaller or have an unusual shape.

5. Skeletal Anomalies

- Spinal irregularities: Some individuals with hemifacial microsomia may have irregularities in the spine, such as vertebral fusion or scoliosis.
- Bony fusion: The bones of the skull or spine may sometimes be fused or misshapen.

6. Hearing Impairment

- Conductive hearing loss: Due to microtia, atresia, or other ear canal irregularities, individuals with hemifacial microsomia may experience hearing loss. This is often conductive hearing loss, which occurs when sound waves cannot be transmitted efficiently through the outer or middle ear.

7. Nervous System Involvement

- Facial nerve weakness: In some cases, individuals with hemifacial microsomia may have weakness or paralysis of the facial muscles on the affected side due to facial nerve involvement.

8. Other Features

- Skin tags: Small, benign growths of skin may appear near the ears or other areas of the face.
- Cardiac anomalies: Some individuals with hemifacial microsomia may also have heart difficulties, although this is less common.

The severity of hemifacial microsomia can vary widely, from mild cosmetic issues to more severe structural differences and functional difficulties.

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Causes

The exact cause of hemifacial microsomia is often unknown, but several factors have been identified that may contribute to its development.

1. Genetic Factors

- Genetic mutations or syndromes: In some cases, hemifacial microsomia may be linked to genetic mutations or inherited syndromes. In these cases, the syndrome is usually caused by a mutation in specific genes that affect craniofacial development.
- Inheritance: In some cases, hemifacial microsomia is inherited as part of a genetic syndrome (like the ones mentioned above), while in other cases, it occurs sporadically without any family history. When it is inherited, it typically follows an autosomal dominant pattern, meaning a single copy of the mutated gene is enough to cause the condition.

2. Vascular Disruptions

- Blood flow issues during pregnancy: One of the most widely accepted theories for the development of hemifacial microsomia is vascular disruption during foetal development. It is thought that problems with blood flow to the developing tissues of the face can lead to underdevelopment of the structures on one side of the face, particularly the ear, jaw, and cheekbones.
  - These disruptions may occur during a specific window of time in foetal development when the first and second pharyngeal arches (structures that give rise to parts of the head and neck) are forming.
  - If blood flow is disrupted in one area of the face, the affected side may fail to develop properly, leading to the typical features of hemifacial microsomia.

3. Spontaneous Mutations

- Sporadic occurrence: In many cases, hemifacial microsomia occurs sporadically, meaning there is no clear family history or environmental exposure that could explain the condition. In these cases, the cause may be related to random mutations or disruptions during early development, though the exact mechanisms are not fully understood.

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4. Alterations in Early Developmental Processes

- Embryonic development: Hemifacial microsomia is thought to occur during the early stages of embryonic development, specifically around the fourth to eighth weeks of gestation, when the first and second branchial arches (structures that contribute to the formation of the face) are developing. Interruption of the normal development of these structures can lead to incomplete or asymmetric facial development.

In many cases, hemifacial microsomia arises sporadically, meaning there may be no identifiable cause, although the underlying genetic, environmental, or developmental factors remain important in understanding the condition.

Testing & Diagnosis

The testing and diagnosis of hemifacial microsomia typically involves a combination of clinical evaluation, imaging studies, and sometimes genetic testing. The condition can often be identified shortly after birth, based on the distinctive facial features, but additional tests are performed to confirm the diagnosis and assess the severity of the condition.

1. Clinical Evaluation

- Physical examination: The first step in diagnosing hemifacial microsomia is a detailed physical exam, where a doctor will assess the appearance of the face, ears, and other related features. This includes looking for:
  - Facial asymmetry: The degree of underdevelopment on one side of the face.
  - Ear anomalies: Such as microtia (underdeveloped ear) or aural atresia (absence of the ear canal).
  - Jaw and dental irregularities: A smaller jaw, cleft lip/palate, or missing teeth may also be noted.
  - Eye anomalies: Colobomas (variations in eye formation where a gap or notch occurs in structures like the iris, retina, or optic nerve) or other eye differences.
  - Other related conditions: The doctor will look for other problems, such as spinal issues, heart problems, or skeletal anomalies, which may suggest a genetic syndrome associated with hemifacial microsomia.

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- Family history: The doctor may ask about any family history of similar conditions to see if hemifacial microsomia could be inherited as part of a syndrome.

2. Imaging Studies

Imaging helps assess the extent of any problems, especially those affecting the bones, joints, and ears, and can provide insight into the degree of asymmetry.

- X-rays: These may be used to assess bone development, especially in the jaw and skull. X-rays can also help detect vertebral anomalies or other skeletal issues associated with hemifacial microsomia.
- CT scan (Computed Tomography): A CT scan is a more detailed imaging technique often used to assess the craniofacial bones, especially the jaw, cheekbones, and skull, as well as ear structures. It can give a clearer view of bone irregularities such as microtia (outer ear is underdeveloped) or atresia (absence of the ear canal).
- MRI (Magnetic Resonance Imaging): An MRI may be used to assess soft tissue structures and any irregularities in the spinal cord or brain (if there is suspicion of associated neurological issues). MRI is also useful for visualizing muscle development and facial nerve function.
- Ultrasound: An ultrasound may sometimes be used during pregnancy to detect facial differences in the foetus, although this is less common for diagnosing hemifacial microsomia.

3. Genetic Testing

While hemifacial microsomia is primarily diagnosed through clinical evaluation and imaging, genetic testing may be recommended if the condition is suspected to be part of a syndrome or other inherited condition. This can help identify any underlying genetic causes.

Genetic testing may involve:

- Chromosomal analysis: A karyotype or chromosome microarray may be done to check for large-scale chromosomal anomalies or specific mutations associated with craniofacial syndromes.
- Targeted gene testing: If a specific genetic syndrome is suspected, doctors may perform gene testing to look for mutations in known craniofacial development genes.

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4. Audiological Evaluation

Since hemifacial microsomia can lead to hearing loss, particularly in cases with ear anomalies such as microtia or atresia, a hearing test is often performed. This could involve:

- Otoacoustic emissions (OAE): To measure the function of the inner ear.
- Auditory brainstem response (ABR): To assess how the brain processes sounds.
- Tympanometry: To evaluate the function of the middle ear.

5. Prenatal Diagnosis

- In some cases, ultrasound during pregnancy may reveal early signs of hemifacial microsomia, such as facial asymmetry or ear anomalies. If these irregularities are detected, further imaging and testing can be done to assess the extent of the condition and rule out associated complications.

The diagnosis of hemifacial microsomia is often made at birth or shortly thereafter, based on characteristic features. The use of imaging, genetic testing, and audiological evaluations helps to confirm the diagnosis and assess the extent of the condition, guiding further management and treatment.

Treatment

Treatment for hemifacial microsomia depends on the severity of the condition and which structures are affected. The goal is to improve facial symmetry, jaw function, hearing, and overall quality of life.

1. Jaw & Facial Bone Reconstruction

Many children with hemifacial microsomia have underdeveloped jaws (mandibular hypoplasia), leading to asymmetry. Treatments include:

a. Bone Grafts

- Used if the lower jaw is too small in order to reconstruct the joint with rib and cartilage to encourage growth.
- Bone is taken from the rib, hip, or skull and placed in the jaw.
- Typically done in teen years or adulthood when growth has slowed.

b. Orthognathic (Jaw) Surgery

- Done in late adolescence or adulthood after growth is complete.
- Addresses bite issues, facial asymmetry, and jaw misalignment.
- Often combined with orthodontics (braces).

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2. Ear Reconstruction & Hearing Support

Many individuals with HFM have microtia (small or missing ear) and hearing loss due to middle ear anomalies. Treatment options:

a. Ear Reconstruction Surgery

- Autologous Reconstruction: Uses the patient's rib cartilage to sculpt an ear (usually done in stages around ages 6-10).
- Prosthetic Ears: Synthetic implants for a more defined shape, sometimes preferred over rib cartilage reconstruction.

b. Hearing Aids & Cochlear Implants

- If the ear canal is missing or malformed, a bone-anchored hearing aid (BAHA) is recommended.
- If inner ear function is affected, a cochlear implant may help restore hearing.

3. Soft Tissue & Muscle Augmentation

Facial asymmetry is often caused by underdeveloped muscles, nerves, and fat tissue. Treatments:

a. Fat Grafting or Implants

- Adds volume to the cheek, chin, or lips to balance the face.
- Often performed in childhood or early adolescence.

b. Facial Reanimation Surgery

- If facial nerves are weak or absent, a nerve or muscle transplant can restore movement.
- Sometimes involves a gracilis muscle transfer (from the leg).

4. Dental & Orthodontic Treatment

Because hemifacial microsomia affects the jaw and teeth, many patients need orthodontic and dental care:

- Braces or other dental treatments to address misaligned teeth.
- Palate expanders to widen the upper jaw if it is small.
- Prosthetic teeth or implants if teeth are missing.

5. Speech, Physical, and Occupational Therapy

- Speech Therapy: Helps with speech and language development if jaw or tongue movement is affected.
- Physiotherapy: Strengthens facial muscles if weakness is present.
- Occupational Therapy: Helps improve chewing, swallowing, and daily activities.

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When Is Treatment Started?

- Infancy – Early Childhood (0-5 years): hearing aids, speech therapy, early evaluations.
- Childhood (5-10 years): jaw distraction osteogenesis, ear reconstruction begins.
- Adolescence (10-18 years): orthodontics, soft tissue procedures, final jaw surgery.
- Adulthood (18+ years): additional cosmetic refinements, orthognathic surgery if needed.

Did you know?

In Australia, hemifacial microsomia is diagnosed in approximately 6 babies per 100,000 births.*

No two cases of hemifacial microsomia are exactly alike—the condition affects everyone differently, making each person's facial features uniquely asymmetrical in their own special way.

References:

Junaid, M. 2024 personal communication: Prevalence data for craniofacial anomalies in WA between 1983 and 2020.

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How we can support you:

- Care packs
- Financial assistance
- Family support coordinator
- Connection to other families