Goldenhar (Hemifacial Microsomia)

BIRTH DEFECT RESEARCH FOR CHILDREN

What is Goldenhar Syndrome?

Goldenhar Syndrome is a rare birth defect characterized by abnormal prenatal development of the head and face. Common features include missing ears and malformations of the jaw, eye, vertebrae, mouth and palate.

Goldenhar Syndrome

Goldenhar Syndrome is also a disorder with many names. Some doctors differentiate between types with minor details. The following is a list of other names used for

Goldenhar Syndrome:

Facio-Auriculo-Vertebral Spectrum (FAV)
Goldenhar-Gorlin Syndrome
First and Second Branchial Arch Syndrome
Hemifacial Microsomia (HFM) - subdivision
Oculo Auriculo Vertebral Spectrum (OAV)

What causes Goldenhar Syndrome?

Although originally thought to occur randomly, there have been a few cases suggesting family inheritance. Research is currently being done on the possible interaction of some environmental factors on genes, suggesting a multifactorial inheritance. Thalidomide, Primidone, and Accutane produced birth defect patterns that are similar to Goldenhar Syndrome. Also, a high rate of Goldenhar Syndrome has been found in the children of Gulf War veterans. Research on birth defects due to the exposures in a hazardous wartime environment is being conducted.

Symptoms include:

• Abnormalities in the head and facial or bones of the spinal column. The child's face may appear smaller or more asymmetrical on one side than the other. Cheekbones, upper and lower jaw bones, and the bones forming lower skull may be underdeveloped.

• Incomplete development of certain facial muscles. An abnormally wide mouth. Cleft palate and/or cleft lip Individuals may exhibit absence or malformation of the outer ears.

• Abnormalities also may exist in middle/inner ear canals resulting in hearing impairment. Abnormal outgrowths of skin and cartilage around ear area.

• Eye abnormalities including cysts on the eyeballs and absence of tissue from upper eyelids. Can include absence of eye.

• Incomplete development, fusion, or absence of certain vertebrae.

In addition affected individuals may have:

- Mild retardation
- Psychological and emotional problems.
- Other skeletal, neurological, heart, lung, kidney, and gastrointestinal abnormalities.

How can you help a child with Goldenhar Syndrome?

Diagnosing GS: A genetic or birth defect specialist should be consulted for diagnosis. Also, a high resolution ultrasound may be used for detection of skeletal abnormalities.

Early Identification: Audiological evaluation at an early age will help to detect and manage a hearing loss. Even babies with profound hearing loss usually have some residual hearing. This residual hearing can often be improved to a great extent through the use of a hearing aid and special training. It is also important for intervention in helping these children to develop speech skills that may be lost if they do not receive help until later. A speech pathologist/ hearing specialist can offer therapy to help a child succeed in learning the basic skills.

Surgery: A baby's cleft lip and/or palate can be repaired surgically. Timing of these surgeries will depend on the health and weight of the baby, but generally the sooner the better. Many babies with oral clefts will have difficulty nursing or drinking from a bottle. It may take extra time, but it will become easier as parents learn to adjust to their baby's special needs. Plastic surgery may improve the facial appearance which in turn can help improve any psychological and emotional problems.

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Dentistry: Dental care may also be required to correct any anomalies. Pediatric dental specialists should be consulted as early as possible to make sure the child's jaw is the right size and shape to correct the position of individual teeth and to maintain good dental and oral hygiene.

Fact Sheet by:

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