



Cystic Hygroma

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Cystic hygromas are fluid-filled sacs that result from a blockage in the lymphatic system. The lymphatic system is a network of vessels that maintains fluids in the blood, as well as transports fats and immune system cells. Cystic hygromas are single or multiple cysts found mostly in the neck region. A cystic hygroma can be present as a birth defect (congenital) or develop at any time during a person's life. A cystic hygroma in a developing baby can progress to hydrops (an excess amount of fluid in the body) and eventually fetal death. Some cases of congenital cystic hygromas resolve leading to webbed neck, edema (swelling), and a lymphangioma (a benign yellowish-tan tumor on the skin composed of swollen lymph vessels). In other instances the hygroma can progress in size to become larger than the fetus.

Cystic hygromas occur in approximately 1% of fetuses between weeks 9 and 16 of pregnancy.

Causes:

Cystic hygromas can occur as an isolated finding or in association with other birth defect as part of a syndrome. They result from environmental factors, genetic factors, or unknown factors.

Environmental causes for cystic hygroma include:

- Maternal viral infections, such as Parvovirus of Fifth's disease
- Maternal substance abuse, such as abuse of alcohol

Genetic syndromes with cystic hygroma as a clinical feature:

- The majority of prenatally diagnosed cystic hygromas are associated with Turner syndrome, a chromosome abnormality in which a female has only one X chromosome instead of two.
- Chromosome abnormalities such as trisomies 13, 18, and 21
- Noonan syndrome

The pattern of inheritance for these syndromes varies depending upon the specific syndrome. Isolated cystic hygroma can be inherited as an autosomal recessive disorder for which parents are "silent" carriers. Finally, a cystic hygroma can occur from an unknown cause.

Testing:

Because the risk for a chromosome abnormality approaches 50% when a cystic hygroma is found prenatally, amniocentesis or CVS (chorionic villus sampling) can be performed to diagnose these conditions. If a genetic syndrome is suspected, test availability depends on the specific disorder. Maternal viral studies may also be considered if hydrops is present or maternal exposure can be documented.

Work up for a prenatally diagnosed cystic hygroma includes:

- A detailed ultrasound, including fetal echocardiogram, to look for other anomalies that may indicate the cause for the hygroma
- A complete family history to determine if testing is indicated for hereditary syndromes.
- Amniocentesis or CVS to look for chromosome abnormalities or a specific genetic syndrome. Viral studies on amniotic fluid can be performed if indicated by the presence of hydrops. (Maternal serum screening does not help in determining the prognosis for a fetus with a cystic hygroma.)
- Periodic ultrasound evaluations are necessary to look for resolution of the cystic hygroma and/or development of other anomalies or fetal hydrops.

In the event of a fetal demise, a complete post-mortem exam (autopsy) is helpful to determine risks to future pregnancies.

Prognosis:

In some situations, a cystic hygroma can be present in a healthy baby. If a chromosome abnormality is not found in the fetus, the outcome is generally better than for those who do have a chromosome abnormality. If a cystic hygroma is an isolated finding that resolves around 18-20 weeks gestation and the fetus has normal chromosomes, the outcome is good for 54-80% of these cases. In cases in which an isolated cystic hygroma does not resolve by 20 weeks gestation, 2-9% have a good outcome. Overall, there is generally a poor prognosis associated with the prenatal finding of cystic hygroma. Studies have indicated that smaller cystic hygromas are more likely to resolve. Oligohydramnios (not enough amniotic fluid) or polyhydramnios (too much amniotic fluid) predicts a poor outcome. Hydrops occurs 22-76% of the time with a cystic hygroma and is almost always associated with miscarriage or fetal death.

Treatment:

A baby with a prenatally diagnosed cystic hygroma should be delivered in a major medical center equipped to deal with neonatal complications. An obstetrician usually decides the method of delivery. If the cystic hygroma is large, a cesarean section may be performed. After birth, infants with persistent cystic hygroma must be monitored for airway obstruction. A thin needle may be used to reduce the volume of the cystic hygroma to prevent facial deformities and airway obstruction. Close observation of the baby by a neonatologist after birth is recommended. If resolution of the cystic hygroma does not occur before birth, a pediatric surgeon should be consulted.

Cystic hygromas that develop in the third trimester (after thirty weeks gestation) or in the postnatal period are usually not associated with chromosome abnormalities. There is a chance of recurrence after surgical removal of the cystic hygroma. The chance of recurrence depends on the extent of the cystic hygroma and whether the wall of the cyst was able to be completely removed.