Jarcho Levine Patient Information

Patient Information Series – What you should know, what you should ask.

What is Jarcho-Levine Syndrome?
Jarcho-Levine Syndrome is a very rare congenital disorder that causes abnormalities in the way bones in the ribs and spines are formed. This causes shortening of the neck and the torso and can lead to underdevelopment of the lungs due to the small size of the chest. Several subtypes of this condition are recognized, with differing prognoses. The syndrome is also known by several different names including costovertebral segmentation anomalies, spondylocostal dysostosis, spondylocostal dysplasia, spondylothoracic dysostosis, spondylothoracic dysplasia and occipito-facial-cervico-thoracic-abdomino-digital dysplasia.

How does it happen?
Jarcho-Levine Syndrome occurs because of mutations in one of several genes. An unborn baby can inherit this condition when both parents carry a recessive gene for the syndrome. Rarely, it can be inherited when one parent passes on a dominant gene (not recessive) to the offspring. Even though this syndrome is found in several ethnic groups, it is most prevalent in people of Puerto Rican descent.

How are chromosomes relevant to Jarcho-Levine Syndrome?
The genes that have been associated with the inheritance of the syndrome are located on the chromosomes inherited by the unborn baby. When other structural differences are evident in the fetus affecting other systems, a chromosomal abnormality may be present.

Should I have more tests done?
A diagnosis of Jarcho-Levine syndrome is often made after birth however; it can be detected during a routine scan to look for abnormalities in the unborn baby during the antenatal period. Signs that the condition may be present in your baby include a small chest size, poorly formed spine, absent or fused ribs, etc. Magnetic resonance imaging (MRI) may be used to support the diagnosis. When there is a suspicion following an antenatal fetal ultrasound, molecular genetic testing should be done to identify mutations in the genes responsible for Jarcho-Levine Syndrome, or other chromosomal abnormalities. A detailed ultrasound examination will be necessary to rule out other structural differences in the baby that can influence outcome.

What are the things to watch for during pregnancy?
Your doctor may advise additional ultrasound exams to assess the interval growth and well-being of the baby. From a maternal standpoint, you can expect that follow up during the pregnancy will follow the routine standard of care for normal pregnancies.

What does it mean for my baby after it is born?
Your baby must be delivered in a center with facilities to manage babies with complex medical problems. The subtype of the condition ultimately affects the outcome of the baby: the small size of the chest, condition of the lungs and the presence or absence of other structural differences. . The baby is likely to require resuscitation in the delivery room because of pulmonary hypoplasia (poorly developed lungs). Within the first few months of life, just under half of the newborns affected by this condition die because of lung underdevelopment and its attendant complications including recurrent lung infections. Surgery is sometimes required. Longer-term survival into adulthood has been reported in some cases.

Will it happen again?
There is a 1-in-4 chance of the conditioning recurring in each subsequent pregnancy.

What other questions should I ask?

- What other potential problems can my baby have?
- What are the long-term survival changes of my baby?
- Is termination of pregnancy an option?
- Can my baby's survival chances be improved?
- Will my baby have normal neurological development?

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