Short Rib Polydactyly syndrome

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

What is Short Rib Polydactyly syndrome?

Short rib polydactyly syndrome (SRPS) is a rare (2.5-3.3 per 10,000 births) group of lethal skeletal anomalies characterized by hypoplastic (very small) thorax, short ribs, short limbs, polydactyly (additional fingers), and visceral abnormalities. Although frequently present, polydactyly may be missing.

How does short rib polydactyly syndrome happen?

SRPS is considered as part of a wide group of genetic skeletal abnormalities, which have been associated to gene mutations. SRPSs are considered on the most severe end of the spectrum, being lethal conditions in most of the cases, while the other skeletal ciliopathies are not uniformly lethal.

How is it diagnosed?

Prenatal diagnosis can be suspected on ultrasound by detecting the following characteristic triad:

- Micromelia (short limbs)
- Short and horizontal ribs with narrow thorax and
- Polydactyly (not constant finding).

Increased nuchal translucency or nuchal fold, generalized edema, ascites (build-up of fluid in the space between the lining of the abdomen and abdominal organs) or hydrops (accumulation of fluid in more than one part of the body) are often observed in cases of SRPSs.

How is it classified?

There are four types of SRPS, including

- Saldino Noonan syndrome (SRPS type I) is the most rare and severe form of SRPS. In addition to the characteristic features of SRPSs, limbs are severely shortened, with absent or hypoplastic fibulae. At the level of the spine very small vertebrae and iliac bones is characteristic. Congenital heart malformations, cleft lip and palate, gastrointestinal, renal and genital anomalies have been associated with this syndrome
- Majewski syndrome (SRPS type II) is characterized by short ovoid tibias (shorter than fibulae), sometimes absent tibial, in addition to the typical short ribs and short limbs (micromelia or mesomelia). Additional anomalies affecting the airway, lungs, kidneys, gastrointestinal, genitourinary and brain can also be present.
- Verma-Naumoff syndrome (SRPS type III) is the most common type and is considered a milder form of type I SRPS, characterized by the typical SRPSs features (severely

short ribs and limbs, inconstant polydactyly). Also, different anomalies such as cleft lip, or kidney, gastrointestinal, urogenital, brain and/or cardiac malformations can be present.

- Beemer-Langer syndrome (SRPS type IV) is clinically similar to SRPS type II, characterized by short ribs, short limbs, but less small tibiae, with bowed long bones, especially in the upper limbs. Polydactyly is rarely present in this variant. Cleft lip and/or palate, hydrops, ascites, macrocephaly and brain abnormalities are also typical findings, even if less frequent than in SRPS type II.
- A new type V SRPS has been suggested, similar to type III SRPS with additional bone abnormalities

Should I have any more tests?

Evaluation should be carried out by an expert in Fetal Medicine. Additional sonographic (by ultrasound) evaluation should include a detailed exam of all other organs, as additional anomalies that may also impact prognosis are usually present.

What is the prognosis?

While SRPS I and II are considered severely lethal during the perinatal period, SRPS type III can be considered a milder variant, with fewer visceral anomalies.

Will it happen again?

SRPS has an autosomal recessive pattern of inheritance (both parents must be carriers) with a 25% risk of recurrence.

What other questions should I ask?

- Should I have any more tests?
- Are there any additional anomalies present?
- Have other skeletal anomalies been ruled out?

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