Potter’s Syndrome — Causes and Prognosis

Potter syndrome, also known as the Potter sequence refers to the specific appearance of the baby due to oligohydramnios a decrease in amniotic fluid volume that leads to deformities in the fetus while in the uterus. These deformities include flattened nasal bridge, epicanthal folds, low set ears and bowed legs. It also leads to pulmonary hypoplasia. The predominant cause is the low production of amniotic fluid which is required to cushion the baby against the uterine walls.

Causes of Potter´s Syndrome

The primary problem in potter syndrome is the failure of the kidneys to develop properly as fetus kidneys contribute to produce amniotic fluid in the form of urine. Approximately after 16 weeks of gestation, fetus excrete urine which determines the volume of the amniotic fluid. The fetus continuously swallows the urine which gets reabsorbed by the gastrointestinal tract and is secreted by the kidneys.

If the kidneys of the fetus are not functioning properly, there will be decreased production of urine which means there will be a decreased volume of amniotic fluid to cushion the fetus against the uterine walls. To date, there is no known genetic cause leading to potter syndrome.

There are other known genetic abnormalities which lead to potter syndrome
and these are listed below:

- Autosomal dominant polycystic kidney disease
- Autosomal recessive polycystic kidney disease
- Hereditary renal dysplasia
- Bilateral Renal Agenesis or BRA
- Some anomalies of the cardiovascular, skeletal, and central nervous system

Other causes include atresia of urethra or ureter, toxemia, amniotic rupture and uteroplacental insufficiency due to maternal hypertension. The classic form of Potter sequence occurs with **agenesis of ureters due to bilateral renal agenesis**. Recent researches have shown the condition to occur twice in males as compared to females which shows the disease association to be linked to Y chromosome but till now no known specific gene has been identified to cause the disease.

**Signs and Symptoms of Potter’s Syndrome**

In cases of bilateral renal agenesis or unilateral agenesis, the metanephros will fail to develop which causes degeneration of metanephros. The mesonephric ducts will also degenerate and fail to connect to the bladder. Therefore, there will be compromised or failure of production of urine and its transportation into the bladder which causes decreased production of amniotic fluid. The decreased amount of amniotic fluid will not be able to cushion the baby against the uterine walls leading to compression and deformities in the fetus.

**Following features will be present in the fetus with Potter syndrome:**

- Flattened nose
- Recessed chin
- Epicanthal folds (skin fold extending from the medial canthus and going across the cheek)
- Low set abnormal ears
- Pulmonary hypoplasia
- Skeletal abnormalities
- Clubbed feet
- Bowing of legs
- Diaphragmatic hernia
- Eye anomalies like cataract, prolapse of lens
- Cardiovascular abnormalities such as ventricular septal defect, Patent ductus arteriosus and tetralogy of Fallot

**Types of Potter’s Syndrome**

Potter sequence has been classified into five distinct types which are as follows:

<table>
<thead>
<tr>
<th>Form</th>
<th>Cause</th>
</tr>
</thead>
<tbody>
<tr>
<td>Classic form</td>
<td>Bilateral renal agenesis</td>
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<tr>
<td>Type 1</td>
<td>Autosomal recessive polycystic kidney disease</td>
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<tr>
<td>Type 2</td>
<td>Renal agenesis</td>
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<tr>
<td>Type 3</td>
<td>Autosomal dominant polycystic kidney disease</td>
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<tr>
<td>Type 4</td>
<td>Obstruction in ureters or kidneys or hydronephrosis</td>
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Prognosis of Potter’s Syndrome

The prognosis is poor for the patients with Potter syndrome. Most babies are still born. Others usually survive approximately until 8 years of age and ultimately need a kidney transplant. Those who survive have chronic kidney disease and require dialysis regularly.

References


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