Potter Syndrome — Causes and Prognosis

See online here

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

Causes of Potter Syndrome

The primary condition associated with Potter syndrome is **failure of the kidneys to develop properly,** as fetal kidneys contribute to produce amniotic fluid in the form of urine. At approximately 16 weeks' gestation, the fetus excretes urine, which determines the volume of amniotic fluid. The fetus continuously swallows the urine, which is reabsorbed by the gastrointestinal tract and secreted by the kidneys. If the fetal kidneys are not functioning properly, there will be decreased urine production, resulting in a decreased volume of amniotic fluid to cushion the fetus in the uterus. To date, there is **no known genetic cause** leading to Potter syndrome.

There are other known genetic abnormalities that lead to Potter syndrome, which are listed below:

- Autosomal dominant polycystic kidney disease
- Autosomal recessive polycystic kidney disease
- Hereditary renal dysplasia
- Bilateral renal agenesis (BRA)
- Anomalies of the cardiovascular, skeletal, and central nervous system

Other causes include atresia of the urethra or ureter, toxemia, amniotic rupture, and uteroplacental insufficiency due to maternal hypertension. The classic pathogenesis of Potter sequence occurs with **agenesis of the ureters due to bilateral renal agenesis.** Recent research has shown the condition is twice as likely to occur in males as compared with females. This suggests a disease association that is linked to the Y chromosome, however, no known specific gene has been identified as the cause of the disease.

Signs and Symptoms of Potter Syndrome

In cases of bilateral renal agenesis or unilateral agenesis, the metanephros will fail to develop, which **causes degeneration of the metanephros.** The mesonephric ducts will also degenerate and fail to connect to the bladder. This results in a compromised or failed production of urine and its subsequent transportation into the bladder, resulting in a decreased production of amniotic fluid. The decreased volume of amniotic fluid will not adequately cushion the fetus against the uterine wall, leading to compression and deformities in the infant.

Features associated in a fetus with Potter syndrome:

- Flattened nose
- Recessed chin
- Epicanthal folds (skin folds extend from the medial canthus to across the cheek)
- Low-set abnormal ears
- Pulmonary hypoplasia
- Skeletal abnormalities
- Clubbed feet
- Bowing of legs
- Diaphragmatic hernia
- Eye anomalies (e.g., cataract, prolapse of the lens)
- Cardiovascular abnormalities (e.g., ventricular septal defect, patent ductus arteriosus, tetralogy of Fallot)

Types of Potter Syndrome

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

Form	Cause
Classic form	Bilateral renal agenesis
Type 1	Autosomal recessive polycystic kidney disease
Type 2	Renal Agenesis
Type 3	Autosomal dominant polycystic kidney disease
Type 4	Obstruction in ureters or kidneys or hydronephrosis

Prognosis of Potter Syndrome

The prognosis for patients with Potter syndrome is poor. Most infants are stillborn. Others typically survive until approximately 8 years of age and then ultimately **require a kidney transplant**. Those who survive have chronic kidney disease and require regular dialysis.

References

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