

Understanding Pfeiffer Syndrome

Pfeiffer syndrome occurs in one out of 100,000 newborns.

1 Introduction

Pfeiffer syndrome is a rare birth defect that affects the development of the bones in the skull, hands and feet. This syndrome causes the joints between the skull bones to fuse together too early in development, affecting the shape of the head and face. The hands and feet can also be affected with children often having broad, short thumbs and big toes as well as webbing between the fingers and toes. Pfeiffer syndrome can be caused by genetic mutations during fetal development or passed from parent to child.

2 Types

There are three types of Pfeiffer syndrome:

- **Type 1:** Mildest and most common, this type is characterized by milder symptoms, children have average intelligence and a normal lifespan.
- **Type 2:** This is the most severe type and includes the presence of a cloverleaf deformity caused by a more extensive fusion of

bones in the skull. The child may have significant developmental delays and a shortened lifespan.

- **Type 3:** This type is similar to type 2, but doesn't include the cloverleaf deformity.

3 Symptoms

The symptoms may start to appear during pregnancy and as a newborn:

- **Misshapen head** — wide, high forehead and short head from front to back
- Wide-set, bulging eyes
- Crooked, crowded teeth
- Hearing loss
- **Midfacial hypoplasia** — reduced growth of the middle of the face, causing a sunken facial appearance

4 Treatments

The treatment of Pfeiffer syndrome is individualized, directed toward each child's specific symptoms. Treatment should be addressed immediately after the child is born and typically focuses on surgeries for reshaping the skull and correcting obstructive sleep apnea. More surgeries may occur later in childhood to correct other symptoms of this syndrome.

Did you know?

Fewer than 50,000 people in the U.S. have Pfeiffer syndrome.

For more information on Pfeiffer syndrome and supportive resources, please visit childrensnational.org.

References:

<https://www.chop.edu/conditions-diseases/pfeiffer-syndrome>

<https://rarediseases.org/rare-diseases/pfeiffer-syndrome/#therapies>

<https://www.childrenshospital.org/conditions/pfeiffer-syndrome#:~:text=What%20is%20Pfeiffer%20syndrome%3F,increased%20pressure%20around%20the%20brain.>

<https://rarediseases.info.nih.gov/diseases/7380/pfeiffer-syndrome>