# *Pfeiffer Syndrome*

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## Abstract:

*Pfeiffer syndrome is a rare genetic disorder characterized by premature fusion of certain skull bones (craniosynostosis) and other birth defects in the hands and feet. There are three subtypes of the syndrome, with Types II and III being the most severe. There is no specific treatment for Pfeiffer syndrome. Treatment is directed at improving the individual's symptoms. Pfeiffer syndrome is associated with mutations (changes) in the FGFR genes.It affects about 1 out of every 100,000 children. Pfeiffer syndrome can be inherited or can occur due to a new mutation, or change, in the involved gene. In cases of severe Pfeiffer syndrome, a new mutation is typically the cause.*

***Key words: Pfeiffer syndrome, craniosynstosis, mutations, birth defect .***

## INTRODUCTION

Pfeiffer syndrome is a rare birth defect ,affects the shape of a baby's skull and face.In new born, the top of the skull isn't one solid piece. It's actually made up of several bones with special joints between them. This allows it to expand so the brain has room to grow. Normally, the skull bones come together only after the head reaches it full size. In child with Pfeiffer syndrome, the plates join together too early. The skull can't expand as the brain grows, which affects the shape of the head and face.Pfeiffer syndrome is also referred to as acrocephalosyndactyly type V(ACSV), craniofacial-skeletal-dermatologic syndrome, and Noack syndrome.1

## TYPES:

Pfeiffer syndrome is subdivided into three types.

- Pfeiffer syndrome type I or Classic Pfeiffer syndrome: It includes caraniosynstosis and "midface deficiency" Child with Type I Pfeiffer syndrome usually have a normal lifespan and typical intelligence.
- Pfeiffer syndrome type II The child present with cloverleaf-shaped skull due to extensive fusion of bones as well as proptosis(abnormal protrusion or displacement of an eye or other body part) and child has poor prognosis and severe neurological problems generally with early death.
- Pfeiffer syndrome type III : Those with Types III Pfeiffer syndrome have more severe defects that can impair brain development and functions.3



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**CAUSES**

Pfeiffer syndrome is inherited as an autosomal dominant genetic disorder, which means ,only a single copy of an abnormal gene is necessary to cause the condition. The abnormal gene can be inherited from either parent or be a result of a new mutation in the affected individual2. Almost all cases of Pfeiffer syndrome type II and type III originate from new mutations because the parents are unaffected.

Pfeiffer syndrome type I is associated with mutations in genes known as FGFR1 and FGFR2 Pfeiffer syndrome type II and type III are associated with mutations in FGFR2.

Older age in the father is a known risk factor for acquiring Pfeiffer syndrome in the offspring due to new mutations.

**SIGNS AND SYMPTOMS:**

- Bulging eyes
- Wide-set eyes
- High forehead
- Beaked nose
- Underdevelopment of the upper jaw
- Prominent lower jaw
- Protrusion of the eyes
- Hearing loss (in over 50% of those affected)
- Short fingers and toes (brachydactyly)
- Webbing or fusion between the digits (syndactyly)
- Dental problems
- Wide thumbs and big toes that bend away from the other digits
- Cloverleaf-shaped head (in Type II Pfeiffer syndrome)[1]

**Diagnosis:**

- Family History/Pedigree
- Physical Exam/History
- X-rays
- CT scan
- Genetic tests

**Other disorders are related to pfeiffer syndrome**

There is a spectrum of disorders associated with changes to the FGFR genes that results in facial defects. These conditions include the following

- Apert syndrome-A genetic disorder characterized by premature fusion of certain skull bone.
- Crouzon syndrome-Affects the first brachial arch which is the precursor for formation of maxilla and mandible bone
- Beare-Stevenson syndrome- A genetic disorder characterized by premature fusion of certain skull bone along with a skin abnormality called cutis gyrata(furrow and wrinkled appearance of the skin)
- FGFR2-related isolated coronal synostosis— Uni or bi coronal carniosyntosis
- Jackson-Weiss syndrome-Characterized by craniosynostosis and foot abnormalities which is the most consistent feature
- Crouzon syndrome with acanthosisnigricans (AN)-Craniosynostosis with skin abnormality characterized by thick ,dark,velvety skin in bodyfolds and creases,including the neck and underarms.
- Muenke syndrome-FGFR3 related craniosynostosis.[4]

**TREATMENT:**

There is no treatment that can reverse Pfeiffer syndrome, but treatments are available that address each individual's specific symptoms. Treatments may be needed from a variety of specialists, including surgeons, pediatricians, otolaryngologists (ENT specialists), neurologists, or others. Reconstructive surgeries can help overcome some of the physical defects associated with the syndrome. Surgery tends to play a key role in treatment.

**Skull surgery:** Most children have the first surgery to reshape their skull before 18 months old.For children under 3 months, this can be done with very small openings in the head. For older children, doctors use traditional surgery. Typically, children will need two to four skull surgeries over the course of their lives.

- **Midface surgery:** Some children may need it to correct problems in their jaw and bring their midface bones forward. Children are usually at least 6 years before this surgery is done. Midface surgery can help remove blockages.surgeon can remove the tonsils or adenoids .
- Some children have blockages that make it hard for them to breathe. This leads to sleep apnea. Wearing a "continuous positive airway pressure mask" --CPAP for short is a standard treatment for sleep apnea.

For severe cases, tracheostomy is done for children too young to have other surgeries to correct breathing problem.[5]

**Reference:**


- Robin NH, Falk,MJ;Haldeman-Englert CR, Eagle KA(2007);FGRF-Related cranisynostosissyndromes.GeneReviews.NCBI.PMID 20301628,
- Cohen ,M.M.Jr( 1993);Pfeiffer syndrome update,clinical subtypes and guidelines for differential diagnosis.Am J.Med.Genet.45,300-307
- National Institutes of health ,Genetic and rare Disease (GARD)Infromation Centre(2016);Pfeiffer syndrome :Symptoms
- Stevens CA,Roeder ER,Ser351Cys(2006);Mutation in the fibroblast growth factor receptor 2 gene results in severe Pfeiffer syndrome.Clin Dysmorpho;l;15:187-8
- National Organization for Rare Disorders. "Pfeiffer syndrome." Pub 2015.


## *Answer Key for the Month of Jan – April 2017*

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