

a guide to understanding
pfeiffer
syndrome

a publication of children's craniofacial association

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this parent's guide to Pfeiffer syndrome is designed to answer questions that are frequently asked by parents of a child with Pfeiffer syndrome. It is intended to provide a clearer understanding of the condition for patients, parents and others.

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This booklet is intended for information purposes only. It is not a recommendation for treatment. Decisions for treatment should be based on mutual agreement with the craniofacial team. Possible complications should be discussed with the physician prior to and throughout treatment.

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what is pfeiffer syndrome?

pfeiffer syndrome is a rare disorder associated with premature fusion of the skull sutures (craniosynostosis), broad and deviated thumbs and big toes and partially webbed fingers and toes (syndactyly). It affects approximately 1 in 100,000 individuals.

what causes pfeiffer syndrome?

there is no link between what the mother did or did not do during the pregnancy. Instead, Pfeiffer syndrome is caused by a mutation (change) in the gene for the fibroblast growth factor receptor (FGFR 1 or 2). The FGFR genes play an important role in signaling a cell to divide or mature. A malfunction of this gene may therefore cause premature fusion of the bones of the skull, fingers or toes. Some studies show that Pfeiffer syndrome occurs more often in children with older fathers.

if I have pfeiffer syndrome what are the odds of passing it to my children?

pfeiffer syndrome is a rare, autosomal dominant disorder, meaning it requires only one parent to have the gene to pass it on to the offspring. A parent with Pfeiffer syndrome has a 50% chance of having a child with the disorder.

how do I recognize this condition in my own child?

the major characteristics of Pfeiffer syndrome include craniosynostosis, along with short, broad thumbs and toes.

Craniosynostosis is a process of premature fusion of the fibrous joints (soft spots) of the bones of the skull. In an unaffected child, the skull evenly expands as the brain grows. In a child with craniosynostosis, one or two of the sutures may prematurely fuse, causing abnormal and asymmetric growth of the skull and face. Advanced craniosynostosis or craniosynostosis involving multiple sutures, may cause a build-up of pressure within the skull or a constriction of the growth of the brain. This may result in developmental delays, mental retardation, seizures or blindness. The sutures most commonly involved in Pfeiffer syndrome include the coronal, lambdoid and sagittal sutures.

Patients with Pfeiffer Syndrome have a disproportionately wide head with a high forehead and retruded or sunken mid-face (the area of the face from the middle of the eye socket to the upper jaw). The nose is frequently small with a low nasal bridge. The eyes can be widely spaced (hypertelorism) and prominent (proptotic) because of the shallow eye sockets (orbits).

Approximately 50% of children with Pfeiffer syndrome have some form of hearing loss secondary to an abnormally small ear canal and middle ear. Dental problems are also common. Visual problems can occur because of the position of the eyes or increased intracranial pressure from the premature fusion of the cranial sutures.

Broad, short thumbs and big toes are commonly seen in people with Pfeiffer syndrome. The thumbs and big toes are frequently deviated away from the rest of the digits. Syndactyly (webbing) may occur between the second and third fingers and toes. Unusually short fingers and toes (brachydactyly) can be present.

what types of pfeiffer syndrome are there?

pfeiffer syndrome has been divided into three subtypes based on the severity of symptoms:

Type 1 – Individuals with type 1 Pfeiffer syndrome have premature fusion of the cranial sutures, recessed cheekbones, and finger and toe abnormalities. Neurologic development and intellect are usually normal. Hydrocephalus (build up of fluid around the brain) and hearing loss can occur.

Type 2 – Individuals with type 2 Pfeiffer syndrome have a “cloverleaf” shaped skull deformity because of extensive fusion of the cranial sutures. Proptosis (abnormal protrusion) of the eyes, finger and toe abnormalities, and fusion of the elbow and knee joints (ankylosis) are also common. The cloverleaf skull can cause limited brain growth and mental retardation. The proptosis of the eyes can cause severe visual problems. Many of these individuals have developmental delay and mental retardation.

Type 3 – These individuals have similar features to type 2 Pfeiffer Syndrome, however they do not have the cloverleaf skull.

how is pfeiffer syndrome diagnosed?

the diagnosis of Pfeiffer Syndrome is made on the presence of premature fusion of the cranial bones and broad, short thumbs and first toes. Other syndromes considered when making the diagnosis include Apert, Crouzon, Saethre-Chotzen and Jackson-Weiss.

It is difficult to make the diagnosis on a prenatal ultrasound, because the clinical features of Pfeiffer syndrome vary considerably.

what treatment is available for pfeiffer syndrome?

the care of an infant with Pfeiffer syndrome begins at birth with accurate diagnosis, identification of the child's needs and the location of a proper treatment center. The care of children with Pfeiffer syndrome can involve multiple, complex surgeries which are best managed by a multidisciplinary craniofacial team. This team is composed of a neurosurgeon, plastic surgeon, dentist, orthodontist, audiologist, speech pathologist, otolaryngologist, geneticist and pediatrician. Team members will work closely with you and your child to determine the best treatment plan.

Early surgery to release the prematurely fused skull sutures may be recommended within the first year of life. Releasing the sutures permits expansion for more normal brain and skull growth. The eye sockets can be enlarged during the same operation to help preserve vision. The midface can be advanced at a later age to help improve the individual's

appearance, increase the orbital volume and establish a more normal upper and lower jaw relationship.

Other treatments may include:

- Hearing tests performed early in life to determine whether ear surgery will be required to help preserve hearing,
- Consultation with a dentist during the second year of life,
- Additionally, your team may recommend hand surgery to release webbed fingers.

Your treatment team will discuss the details of these surgeries with you.

how can children's craniofacial association (cca) benefit my family?

CCA understands that when one family member has a craniofacial condition, each person in the family is affected. We provide programs and services designed to address these needs. A detailed list of CCA's programs and services may be found on our website at www.ccakids.org or call us at 800.535.3643.



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