

Pfeiffer Syndrome

Pfeiffer syndrome is a craniosynostosis syndrome resulting from premature fusion of the sutures of the skull resulting in skull deformity. Abnormal skull growth, which results in a pointed or conical head, is also responsible for underdevelopment of the mid-face (upper jaw bone), high arched palate and prominent lower jaw are characteristic. Eyes are wide set and bulge. Teeth erupt in improper positions. Mild hearing loss due to a defect in the middle ear may be present. Thumbs are short and broad; toes are oversized. Hands and feet may be webbed.

It only takes one parent to carry the gene responsible for Pfeiffer syndrome in order for the condition to be passed on to the child. The risk of transmitting the condition is 50% of each pregnancy.