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# Apert Syndrome

Normally, most babies are born with a soft spot that is caused by the baby's skull not being fully developed. While most people might believe that evolutionarily, babies are disadvantaged for having such a vulnerable spot on their body that could kill them if pressed, having a soft spot is actually preferable to being born with a fully formed skull. The reason for the soft spot is so that your brain has room to develop before the skull fuses. The FGF2 gene (fibroblast growth factor receptor 2) found in chromosome 10 is responsible for bone growth. One mutation in the FGFR2 gene is called Apert Syndrome where the skull bones fuse prematurely.

Apert Syndrome, also known as Acrophalo-syndactyly type 1, occurs when the FGFR2 gene mutates and bones begin to ossify, or fuse, prematurely causing babies to be born with fully developed skulls leaving no room for growth. The mutation arises due to missense on either the 252nd or 253rd amino acids that are responsible for the development of bones when mutated results in osteoblasts to begin ossification prematurely as well as excessive FGF signaling. This not only causes early fusion of the skull, but fusion of the fingers or toes called syndactyly. Typically, syndactyly can be seen as the three digits being fused together, and in its most severe

form you can see rosebudding where all fingers are fused. Apert syndrome can also result in polydactyly where there are additional fingers or toes as well.

Apert syndrome is an autosomal dominant disorder and it impacts 1 in 65,000 to 80,000 infants. While the mutation is dominant, the mutation can be de novo or completely new and not inherited from parents. The mutation can either be inherited or it can occur from the paternal age effect where, as the father gets older, sperm begin to mutate. Some symptoms of Apert Syndrome are fusing of the fingers, flattened skull, bulging eyes, mental impairments due to elevated intracranial pressure, sleep apnea, wide-set eyes, sunken middle face, beaked nose, narrow jaw, or cleft palate. While there is no cure for Apert Syndrome there are surgeries that can assist with brain growth, proptosis, and cosmetics. Surgeries can be performed to assist with syndactyly to separate fused fingers. There are also surgeries such as posterior cranial vault distraction where the back of the skull is pushed back to allow for brain growth, fronto-orbital advancement where the front of the skull is pushed forward for brain growth, and Le Fort III osteotomy where the midface is pulled forward.

Apert syndrome can usually be detected in ultrasounds so rarely are parents surprised by Apert Syndrome. Because of this, parents can seek genetic counseling to find ways to make their child's life more comfortable. Genetic counseling can be useful for parents who may have a child with Apert Syndrome because it is typically a de novo mutation and the parents may not have much experience with the disease. However, genetic counseling should not be made mandatory because of a myriad of reasons. One reason is that healthcare is extremely expensive and parents should not be forced to pay for genetic counseling if they do not want to. Another reason is because forcing people to be in genetic counseling enters a territory that is very close to

eugenics. Forcing people with genetic disorders to take mandatory genetic counseling suggests that people with these disorders should not reproduce or that they must take extra precautions with their children that people without genetic disorders do not have to. Mandatory genetic counseling for people with genetic diseases implies that people without genetic diseases will produce children who do not have these diseases. Apert syndrome is an example of how mutations can occur without inheritance and how unpredictable mutations can be.

Works Cited

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