Diprosopus (Craniofacial Duplication) [1]

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Diprosopus [4] is a congenital defect also known as craniofacial duplication. The exact description of diprosopus refers to a fetus with a single trunk, normal limbs, and facial features that are duplicated to a certain degree. A less severe instance is when the fetus [5] has a duplicated nose and the eyes are spaced far apart. In the most extreme instances, the entire face is duplicated, hence the name diprosopus, which is Greek for two-faced. Fetuses with diprosopus often also lack brains (anencephaly), have neural tube [6] defects, or heart malformations. In some cases, if the brain is formed, it may have duplicated structures. Most infants with diprosopus are stillborn and there are fewer than fifty cases documented since 1864.

Researchers have proposed several mechanisms to explain the phenomenon of craniofacial duplication. When two completed identical faces are present, researchers often class diprosopus as a rare variant of conjoined twinning. They consider the two faces to result from cranial bifurcation of the notochord [7] during neurulation [8]. The bifurcation causes two vertebral axes and neural plates to develop alongside each other, complete with neural crest [9] derivatives. Only 0.4 percent of conjoined twins [10] have diprosopus.

Another possible cause for the anomaly is an increase in the expression of the protein Sonic hedgehog (SHH), which is essential for craniofacial patterning during development. In experiments on chicks, researchers replicated many of the phenotypes associated with craniofacial duplication by exposing the embryos to an excess of SHH. In those experiments, chicks were born with two beaks and the eyes spaced far apart. Too little SHH can also affect midline facial structuring, and cause the developing eyes to fuse together (cyclopia [11]).

Diprosopus [4] can be discovered in utero using technology such as ultrasound [12], computer tomography (CT) scanning, magnetic resonance imaging [13] (MRI), and MR angiography. One of the first indications of craniofacial duplication is polyhydramnios, a condition in which there is an abnormally high amount of amniotic fluid present within the amniotic sac [14]. No treatment exists to cure diprosopus, although therapeutic abortion [15] is sometimes an option if the condition is discovered early enough in the pregnancy [16]. Because of the rarity of diprosopus, there are few treatment options or corrective surgery techniques documented.

Duplication of the face and its structures enables embryologists to hypothesize about the mechanism of the duplication. The diverse range of expression involving craniofacial duplication varies so much that it is possible that more than one mechanism may explain the phenomenon. Further examination of such cases may elucidate the mechanisms responsible for diprosopus, as well as the mechanisms responsible for normal development.

Sources


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