

Iniencephaly

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). [Iniencephaly Information Page](#).

Iniencephaly is a rare birth defect caused by improper closure of the neural tube (the part of a human embryo that becomes the brain and spinal cord) during fetal development. Iniencephaly is in the same family of neural tube defects as spina bifida, but it is more severe. In iniencephaly, the defect results in extreme retroflexion (backward bending) of the head combined with severe distortion of the spine. Diagnosis is made immediately after birth because an infant's head is so severely bent backward that the face looks upward. In most infants the neck is absent and the skin of the face is connected directly to the skin of the chest, while the scalp is directly connected to the skin of the back. Most infants with iniencephaly have additional birth defects, such as anencephaly (in which major sections of the brain fail to form), cephalocele (in which part of the cranial contents protrudes from the skull), and cyclopia (in which the two cavities of the eyes fuse into one). Additional birth defects include the lack of a lower jaw bone or a cleft lip and palate. Other parts of the body may be affected, and infants can have cardiovascular disorders, diaphragmatic hernias, and gastrointestinal malformations. For reasons that are still unknown, the disorder is more common among females. No single gene has been identified as the cause for iniencephaly, or any of the neural tube defects. Scientists think these defects have complex causes, mostly likely a mix of genetic and environmental factors.