

POSTER 19

MIDLINE FACIAL MICROSOMIA: A CASE REPORT

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Holoprosencephaly (HPE) is a severe congenital malformation in which the facial skeleton shows aplasia or hypoplasia of the midline bones from the sphenoid and ethmoid down to the premaxillary bone. HPE was originally described in 1963 by DeMyer who, through observation of patients with this condition, established the concept that the face predicts the brain. With the advent of MRI/CT scanning, it has become evident that the face does not always predict the brain in HPE patients. We describe a patient who fits the phenotypic description of HPE, but lacks the brain findings that are typically associated.

The patient is a 9-year-old child with microcephaly, premaxillary agenesis, absence of the columella and nasal septum, flat nasal bones, telecanthus, and no radiographic or chromosomal evidence of holoprosencephaly. Despite having a facial dysmorphology consistent with holoprosencephaly, there is no radiographic or cognitive evidence of a significant brain abnormality. He is a healthy 9 year old boy who is attending regular classes and is excelling at both reading and spelling above his age appropriate level.

This case, along with others in the recent genetics literature, adds to the evidence that the face does not always predict the brain. Such cases raise the question as to whether or not these patients should be classified in the spectrum of HPE when they have a normal “encephalon”. We believe a more accurate description of these patients with midline facial dysmorphology and brains without gross intracranial anomalies would be “midline facial microsomia.”