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Commentary

Demystifying Malformations

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Written records from earliest times contain fascinating, often lurid, descriptions of real or fanciful, grotesque malformations of the human body. Some, purporting to report part human–part animal beings were uncritically accepted, along with documented examples of humans with two heads, horns, or tails. It was not until the first half of the 18th century that physicians attempted to separate the fictional creations of overwrought imaginations from objective observations [1]. Numerous external anomalies were described and were often ascribed to maternal shock during gestation or possibly to an expression of divine wrath or glorification [1].

Documentation of anomalies of all kinds was essentially limited to those that were externally obvious, as systematic dissection of the human body was not done until the 19th century, although sporadic reports of malformed internal organs appeared in the medical literature before that time. These were largely the contributions of anatomists such as Tulp, Morgagni, and Meckel: only later did pathologists add their descriptions of the gross and microscopic features of malformed visceral and nervous system organs.

While this resulted in a plethora of detailed morphological descriptions of a seemingly endless number of anomalies, the majority were just that—descriptive. Attempts to explain the origin on an etiological-embryological basis were (and are) often uncritically accepted, although some were challenged with the result that several different theories were advanced to account for the same malformation.

Increasing refinements in genetic analyses have allowed association of one or more malformations with specific chromosomal defects, although even knowledge of such relationships does not necessarily provide insight into the specific aberrant embryological event(s) that is(are) responsible for the defect(s).

Truwit and Barkovich [2] have provided their colleagues a superb lesson in combining morphologic observations (made, in this instance, by radiologic techniques) with a thorough knowledge of neuroembryology to arrive at a lucid concept of pathogenesis of these lesions.

Lipomas within the intracranial cavity have traditionally been grouped with hamartomatous, malformative growths of the CNS, but their frequent association with developmental anomalies, particularly of the corpus callosum, has been documented repeatedly [3–5]. The authors note that intracranial lipomas are neither hamartomas nor true neoplasms but rather congenital malformations. Actually, the collection of fatty tissues belongs in the category of a choristoma; that is, a mass of tissue histologically normal for an organ or part of the body other than the site at which it is located [6]. Evidence suggesting that this aberrant fatty tissue results from abnormal persistence and maldifferentiation of the meninx primitiva is convincing.

A parallel but rarer meningeal dysplasia consisting of striated muscle fibers in subarachnoid space primarily around the pons is also characteristically associated with CNS malformations typically involving posterior fossa structures [7]. In contrast to lipomas, the dysplastic muscle does not form an obvious mass, and can only be diagnosed by microscopic study. Muscle in the leptomeninges is thought to result from dysplastic differentiation of the pleuripotential cells of the neural crest; this population of embryonic cells normally gives rise to striated muscle of the head.

Other investigators of the embryopathogenesis of complex CNS malformations would do well to follow the superb example set by Truwit and Barkovich if they wish to demystify developmental processes that have gone awry.

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This article is a commentary on the preceding article by Truwit and Barkovich.

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