Occult craniocervical dysraphism and skin markers

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DESCRIPTION

Neural tissue and skin are of common ectodermal origin and therefore anomalies occurring during early embryogenesis may lead to combined neuro-skin malformations. Neural tube closure occurs during primary neurulation starting from the equivalent of the craniocervical junction and proceeding zip-like in both cephalic and caudal directions.¹ Therefore, any defects of the process may reflect spinal dysraphism with, possibly, combined congenital skin lesions.

Occult spinal dysraphism (not exposed but skin-covered neural tissue), in the vast majority of cases, is localised in the lumbosacral area and less commonly in the cervicothoracic area.^{2 3} This report describes an atypical case of occult craniocervical dysraphism with combined skin lesions/markers.

Routine postnatal examination of a term newborn girl revealed a salmon patch (vascular malformation of capillaries) on the occipital and upper cervical areas, a subcutaneous soft but fixed mass (1.3×1.2×1.0 cm) on the midline at the level of the craniocervical junction, and hair tufts above and below the mass along the midline (figure 1). The differential diagnosis of meningocele/myelocoele, lipoma, dermal cyst, fibroma, or hamartoma was made. Brain and spine MRI revealed a

Figure 2 Fluid density tract (T2 TSE sequence, sagittal view, arrow) protruding through the bony defect.

Figure 3 Subcutaneous cyst/occult occipital meningocele (T2 TSE sequence, axial view, arrow).

Figure 1 Salmon patch, subcutaneous soft mass and hair tufts along the midline of occipital and craniocervical areas.

complex anomaly of the craniocervical junction with a fluid density tract (figure 2) extending from the cisterna magna, and a cerebrospinal fluid space, through a bony defect, to a small subcutaneous cyst (figure 3), providing evidence of an occult occipital meningocele. There was no herniation of the cerebellum or intracranial contents, and no abnormalities of the lower spine.

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Learning points

- ▶ Craniocervical dysraphism represents an anomaly with its origin in early embryogenesis (week 3).
- ▶ Neural tissue and skin are of common ectodermal origin and may therefore present combined neuro-skin malformations.
- ▶ Occult spinal dysraphism with skin markers may be atypically located in the craniocervical area.

Competing interests None declared.

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