Klippel-Feil syndrome revisited: diagnostic pitfalls impacting neurosurgical management*

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Received June 4, 1991/Revised November 18, 1991

Abstract. Klippel-Feil syndrome in its most basic definition includes several anomalous conditions of the cervicomедullary junction and suboccipital region. Pediatric neurosurgeons are often involved in surgical palliation of this syndrome, without realizing how the accompanying anomalies may obfuscate management in the older child. A brief review of the embryology of the rhombencephalon helps to clarify the etiology of some of these symptoms which may cause confusion and, occasionally, inappropriate treatment. Illustrative cases will demonstrate some of these pitfalls. Appropriate early intervention, such as posterior fossa decompression, ventricular shunting, and fundal plication, may help to avoid needless morbidity. The advent of magnetic resonance scanning has helped to clarify the diagnosis and resulted in more appropriate treatment in these cases.

Key words: Klippel-Feil syndrome – Craniocervical abnormalities – Rhombencephalon – Hydrocephalus – Cyclic regurgitation

Anomalous fusion of the cervical vertebrae with an accompanying midline defect [3, 6] is not rare and may result in a variety of misdiagnoses and misdirected treatment. We have all had occasion to participate in discussions involving the emergency room physician, the radiologist, and the orthopedic surgeon revolving about the child with congenital block cervical vertebrae who comes to the emergency room or trauma department with complaints of immobility, pain, and distortion of the upper neck after injury. We know by experience that children with fused cervical vertebrae, with or without craniocervical anomalies, are at risk of injury to the upper cervical cord and medulla from relatively minor trauma. Nagib et al. [7] have reviewed this in detail. They placed the children with Klippel-Feil deformity in three groups, group I being made up of children with unstable fusion patterns, group II of those with craniocervical abnormalities, and group III of those with stenosis of the cervical spinal canal (Table 1).

The group II children are particularly of interest to the pediatric neurosurgeon, due to the many anomalies of the medulla which directly affect the autonomic nervous system and cause clinical symptoms that are often confusing and difficult to treat. The most complex anomalies consist of partial or complete absence of cranial nerve nuclei or tracts.

Embryology

A brief review of the embryology of the rhombencephalon is helpful (Fig. 1). The rhombencephalon during fetal development has a dorsal-ventral orientation around the IV ventricle, with a roof, a base, and lateral alar processes. It has a cranial-caudad orientation that is divided into three major parts: the metencephalon consisting of the analge of the cerebellum and pons, the myelencephalon, which becomes the medulla, and the IV ventricle. The IV ventricle and medulla contain the nuclei and tracts of the lower five cranial nerves (eight through twelve). These nuclei and tracts, along with the surrounding reticular formation, fundamentally control the majority of autonomic function of the viscera [1].

Table 1. Major structural anomalies seen with Klippel-Feil syndrome [7]

<table>
<thead>
<tr>
<th>Group I: Unstable fusion pattern</th>
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<tr>
<td>Group II: Craniocervical abnormalities, e.g. myelodysplasia:</td>
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<tr>
<td>1. Spina bifida aperta</td>
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<td>2. Spina bifida cystica</td>
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<td>3. Myelocele</td>
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<td>4. Myelencephalocele</td>
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<td>5. Syringo-hydromyelia</td>
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<td>6. Chiari malformation</td>
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<td>Group III: Cervical spinal canal stenosis</td>
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* Presented at the XVIII Annual Meeting of the International Society for Pediatric Neurosurgery, Paris 1990
Absence of one or both eighth nerves will lead to deafness, manifested by inattention and dysarthria on the part of the child and interpreted by observers as retardation, speech problems, etc. Loss of the vestibular apparatus will result in gait disorder and cyclic vomiting. Maldevelopment of the ninth nerve or nuclei will result in dysphagia, dysphonia, and loss of taste sensation, as well as tachycardia. Clinically, the child may have cyclic regurgitation, cachexia, dehydration, anorexia, drooling, and various cardiac arrhythmias. Webbing and atrophy of the trapezius and sternocleidomastoid muscle due to loss of the eleventh nerve nuclei causes torticollis, gait abnormalities, and deformities of the upper thoracic muscle and bone; the so-called Sprengel's deformity [2].

Absence of both tenth nerve nuclei is probably incompatible with life; however, structural abnormalities of the vagal nuclei and tracts will cause dysphagia, dysphonia, esophageal and pyloric spasm, and dyspnea. Symptoms of tenth nerve anomalies will include hoarseness, "pseudoa-sthma", drooling, regurgitation, bradycardia, and various cardiac arrhythmias. Webbing and atrophy of the sternocleidomastoid muscle due to loss of the eleventh nerve nuclei causes torticollis, gait abnormalities, and deformities of the upper thoracic muscle and bone; the so-called Sprengel's deformity [2, 7].

Without the hypoglossal medullary apparatus, the child suffers from both dysarthric speech and dysphagia, with an inability to chew and swallow food. Abnormalities of the roof and floor of the IV ventricle will result in hydrocephalus and posterior fossa "cysts," complicated by apnea, bradycardia, and sleep disorders [2].

The pediatric neurosurgeon usually encounters these unfortunate children early in infancy. The presence of suboccipital encephalocele and/or meningomyelecele will precipitate early "urgent" consultation from colleagues pediatrics and neonatology. Closure of the midline defects results in, or is accompanied by, the development of hydrocephalus, which in turn demands ventricular shunting [5]. The unstable fusion or absence of cervical vertebrae may not be appreciated, resulting in untoward complications secondary to anesthesia and surgical positioning [7].

The atrophy and "webbing" of the trapezius and sternocleidomastoid may make subcutaneous placement of the shunt tubing difficult and proper positioning and skin preparation impossible. Correction of the hydrocephalus by shunting may obviate, mitigate, or precipitate apnea, bradycardia, and sleep disturbance [8]. The presence of neck abnormalities and difficulty with positioning can lead directly and indirectly to shunt infection and malfunction [7].

Postoperatively, there can be feeding and nutritional complications necessitating gastrostomy and hyperalimentation, which in turn tend to increase shunt malfunction and infection. The presence of a feeding gastrostomy does not stop the cyclic regurgitation in these children.

Case presentations

To illustrate the management difficulties, I will briefly review three cases of Klippel-Feil deformity type II, which we have followed in our clinic over the last several years.

Case 1 (Fig. 2) is a term male infant, born in October 1981. He was noted to have a large craniocervical myelencephalocele at birth, which was closed that day. A ventriculoperitoneal shunt was placed 9 days after birth to correct the hydrocephalus that developed. The infant was re-admitted at the age of 2 months for revision of the peritoneal end of the shunt. Over the next 8 years he was re-admitted to the hospital on 32 occasions, a rate of once every 2–3 months, with episodes of cyclic hyperemesis, dehydration, failure to thrive, shunt malfunction, shunt infection, seizures, and anorexia. Hospital stays varied from 3 days to 2 months. He had multiple shunt revisions and long-term venous cannulization for hyperalimentation, but managed to avoid feeding gastrostomy.

He is presently (Fig. 3) an ambulatory, dysarthric, moderately retarded youngster, with a functioning ventriculoperitoneal shunt which has not had to be revised for 3 years. He has finally (9 months without hospitalization for dehydration) come into balance from a gastrointestinal point of view. He has significant webbing of his neck, with Sprengel's deformity of one shoulder.

Case 2 is a premature female infant delivered in September 1987. At birth, she was noted to have structural anomalies of the external ear on one side with webbing of the neck musculature, and extreme hydrocephalus. A ventriculostomy was placed first, and subsequently she received a right-sided ventriculoperitoneal shunt. This had to be revised to the left side of her head and neck. A diagnosis of Klippel-Feil syndrome was made during her birth admission. She had seven subsequent admissions over the next few years for seizures, shunt revision, fundal plication, and eventually a posterior fossa decompression, the last for apnea spells. Presently (Fig. 4), she is a severely retarded toddler with a functioning shunt. Her last admission was in March 1991, at which time there was disconnection of the shunt in the neck. She is able presently to take only selected solids by mouth, relying on gastrostomy feedings for the majority of her nutrition.

Case 3 (Fig. 5) is a female term infant born in March 1987. At birth she was noted to have a craniocervical