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CLINICS

Neurological pictures in Paediatric Chiari I malformation

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Abstract The clinical features of Chiari I Malformation (CIM) may be related to the compression of dural and/or neural structures at the craniocervical junction or to the associated syringomyelia. Additionally, patients may exhibit symptoms and signs of associated disorders. CIM is a heterogeneous and multifactorial disorder including congenital and acquired forms; it can also be found as an isolated malformation or in association with many clinical conditions. We analyse the clinical features in a series of 65 children with CIM, focusing on the high frequency of associated clinical disorders. We emphasise the importance of a careful clinical and neurological assessment for a proper diagnosis and a correct management of these patients.

Keywords Tonsillar ectopia · Syringomyelia · Children · Symptoms

Introduction

The Chiari I Malformation (CIM) consists of cerebellar tonsils herniation at least 5 mm below the plane of the foramen magnum. The brainstem may be occasionally involved in this malformation and syringomyelia is frequently associated [1].

The CIM may be asymptomatic or may present with a broad and variable range of symptoms and signs in relation to the compression of dural and/or neural structures at the craniocervical junction or to the associated syringomyelia.

The most common presenting symptom is occipital headache and/or neck pain. Other common symptoms and signs can be divided into three types of presentation: a brainstem syndrome with cardiac and respiratory irregularities and cranial nerve dysfunction including otological disturbances; a cerebellar syndrome with ataxia, tremors, clumsiness, nystagmus and dysarthric speech; a spinal cord syndrome with motor and sensory disturbances especially in the hands, and progressive scoliosis [2, 3].

Additionally, patients may exhibit symptoms and signs of associated disorders.

The CIM is etiologically a heterogeneous and multifactorial disorder comprising congenital and acquired forms. Congenital forms are considered to derive from a mesodermal disorder resulting in an underdevelopment of the posterior cranial fossa with an overcrowding of the normally developed hindbrain [2]. Acquired variants have been described secondary to increased intracranial pressure and chronic hydrocephalus, following cerebrospinal fluid diversion from the spine (lumbo-peritoneal shunting procedures, spontaneous CSF leaks, lumbar punctures), and, more recently, as late complication of early ventriculo-peritoneal or cysto-peritoneal shunting [4].

The CIM can be also found as an isolated malformation or, with high frequency, in association with skeletal abnormalities of the cervical spine and craniovertebral junction (atlas assimilation, Klippel-Feil anomaly, retroversion of the odontoid process…) and skull (some

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syndromic and non-syndromic craniosynostosis); furthermore, it has been reported in association with many clinical conditions (such as Neurofibromatosis type 1, Costello syndrome, idiopathic growth hormone deficiency, achondroplasia…) [3, 5].

A number of outstanding publications have addressed the clinical features of CIM in children and adults [2, 5–10]. Most of the previous studies have collected data from neurosurgical series. Over the past two decades, however, the increasing use of MRI for diagnostic purpose in neurological patients has led to a higher detection of CIM in asymptomatic subjects [11].

The purpose of the present communication is to report the clinical and neurological features of a series of asymptomatic and symptomatic children with CIM, focusing on the high frequency of associated clinical conditions, most of which are genetically determined.

Materials and methods

Between Jan 2008 and Dec 2010, 65 children (35 females and 30 males; age range from 2 months to 17.10 years) with CIM were evaluated in the Department of Paediatric Neuroscience of our institution.

The diagnosis was made by brain MRI in all the patients. Cine-flow and spinal cord MRI were performed in all the children. The clinical evaluation consisted of physical and neurological examination, and cognitive assessment. Neurophysiological investigations (polysomnography, ABRs, SEPs) were also carried out.

When dysmorphic features, growth anomalies, other congenital malformations, cognitive and/or behavioural anomalies were present, an associated clinical disorder was always investigated.

Results

The main clinical symptoms and neuroradiological associated features are shown in Table 1.

The indications for brain MRI are reported in Table 2.

An associated clinical condition was defined in 20 of the 65 patients (30%). Details are shown in Table 3.

Discussion

Our population include more asymptomatic subjects (58.4%) than previous reported series of CIM. It has been reported that 37–57% of paediatric patients and 14–30% of adult patients may be asymptomatic at neuroradiological diagnosis. Little is known about the epidemiology of the CIM. So far the malformation has been identified in 0.9–1% of paediatric patients undergoing MRI scans for various clinical indications [6, 8]. It is likely to be that with

Table 1 Main clinical symptoms and neuroradiological associated features of 65 paediatric cases of CIM

	Number of patients	Percentage (%)
Asymptomatic children	38/65	58.4
Symptomatic children	27/65	41
Symptoms		
Valsalva-induced headache	13/27	48
Neck pain/torticollis	3/27	4.6
Tremor	2/27	7.4
Clumsiness	3/27	11.1
Nystagmus	2/27	7.4
Central apnea	1/27	3.7
Lower cranial nerve dysfunction	3/27	11.1
Vertigo	1/27	3.7
Drop attacks	1/27	3.7
Upper and lower extremities dysesthesia and weakness	6/27	22.2
Surgerical procedures		
Suboccipital decompression	26/65	40
Third ventriculostomy	2/65	3
Neuroradiological associated features		
Skull anomalies	6/65	9.2
Craniocervical junction anomalies	14/65	21.5
Scoliosis	2/65	3
Hydrocephalus	7/65	10.7
Cysto-peritoneal shunting	1/65	1.5
Syringomyelia	15/65	23.1
Lipoma of filum terminale	2/65	3

Some patients exhibit more than one symptom; hence the total does not equal to 27

Table 2 Indications for MRI in 65 paediatric cases of Chiari I Malformation

Indications for MRI	Number of patients	Percentage (%)
Headache/neck pain	26/65	40
Psychomotor/mental retardation	23/65	35.4
Autism	3/65	4.6
Craniosynostosis	4/65	6.1
Cloacal exstrophy	1	1.5
Arachnoid cyst	1/65	1.5
Pseudotumor	1/65	1.5
Neurofibromatosis type 1	1/65	1.5
Epilepsia	2/65	3
Precocious puberty	1/65	1.5
Others	2/65	3

Table 3 Associated clinical conditions in 65 paediatric cases of Chiari I Malformation

Diagnosis	Number of patients
Costello syndrome	2/65
Idiopathic growth hormone deficiency	2/65
PTEN syndrome	1/65
Cutis marmorata telangiectatica congenita	1/65
Charge syndrome	1/65
Alport syndrome	1/65
Angelman syndrome	1/65
Opitz syndrome	1/65
Neurofibromatosis type 1	1/65
Crouzon syndrome	1/65
Muenke syndrome	1/65
Polysynostosis	2/65
Sagitall synostosis	1/65
MEF2C haploinsufficiency	1/65
2p15p16 haploinsufficiency	1/65
TAR syndrome	1/65
Cloacal exstrophy	1/65

the increasing use of MRI in the diagnostic neurological work-up, more and more asymptomatic patients will be identified.

The clinical features of our series are similar to those reported in the previously published series.

The most common presenting symptom in adult and children (60–70%) is occipital headache and/or posterior neck pain [3]. This pain may be dull and persistent, probably in relation to the irritation of the dura of the posterior fossa, but usually it is severe and paroxysmal, induced or exacerbated by Valsalva manoeuvres such as coughing, laughing, sneezing, likely in relation to impaction of the cerebellar tonsils at the level of the foramen magnum with the waves of increased intracranial pressure. In infants or non verbal children, occipital or cervical pain may manifest as irritability or crying or neck arching [3].

In our series, headache represented the most common indication for neuroradiological investigations, as it was referred by 40% of the patients. As headache is a multifactorial condition, we considered it symptomatic for CIM in the 13 children (48%) in whom headache was Valsalva-induced.

The cerebellar, brainstem and spinal cord symptoms and signs in CIM patients classically have an insidious onset. They progress becoming clinically obvious during early adulthood [2, 8].

Rarely, acute spinal cord or bulbar deficits develop with relatively minor head or neck injuries, particularly, in patients with syringomyelia [12]. In one case of acquired CIM secondary to early cysto-peritoneal shunting, we observed acute and dramatic bulbar deficits which required urgent suboccipital decompression surgery; no minor head or neck trauma was reported before symptoms onset.

The clinical manifestations of paediatric and adult patients seem to be similar. However, the more common occurrence of lower cranial nerve dysfunction (up to 77%) has been reported in children younger than 3 years. These patients typically present with poor feeding, failure to thrive, recurrent aspiration pneumonia, dysphagia and central sleep apnea [3, 7, 9]. In our cohort, lower cranial nerve dysfunction and apnea were detected in 14.8% of symptomatic children, all younger than 3 years.

The frequency of other clinical findings is consistent with previously reported series.

Ocular and otological symptoms such as nystagmus, diplopia, tinnitus, fluctuating hear loss, are frequent in adults (74–78%), but more unusual in children (7–19%) [2, 8, 10].

Unusual manifestations (<1%) include hypertension, sinus bradycardia and syncope (Chiari drop attack), probably due to dysfunctional autonomic cardiovascular regulation caused by hindbrain compression [3, 5].

Cerebellar symptoms are less commonly reported than brainstem and/or cranial nerve dysfunction in paediatric age (up to 10%) [8].

When syringomyelia is present, a progressive levoscoliotic spine curve is a frequent sign (16–38%) and may be the first manifestation [9]. Additional symptoms and signs related to the syrinx may be radicular pain and/or dysesthesia, particularly, in the upper limbs with or without numbness and weakness in the hands (6–8%) [5, 8]. In our sample, 15 patients (23.1%) had a syringomyelia; 6 of them developed dysesthesia and weakness of limbs. None presented scoliosis except 2; they were the only patients harbouring a syrinx extended to the whole spinal cord.

During the last decades some studies have focused on a subgroup of CIM patients who also presented language delay, mental retardation with or without epilepsy [13–15]. They suggested that this association may not be coincidental but the expression of subtle cerebral dysgenesis and they speculated a different pathogenetic background compared to CIM patients without neurodevelopmental disorder [13, 14].

It is noteworthy that we found a higher proportion of associated clinical conditions than previously reported, most of which are genetically determined and characterised by neurodevelopmental delay and other neurological deficits (Table 3). Global developmental delay represented a frequent indication for MRI, because it is relatively common in the general population affecting 1–3% of the children [16]. An accurate etiologic determination of developmental delay is mandatory for its implications

regarding treatment, prognosis, medical management and familiar counselling. This is frequently possible in clinical practise, on the basis of the history, physical and neurological examination, neuroradiological and genetic studies. The correct diagnosis of the associated disease may play an important role to ascertain which symptoms are CIM related, so influencing the surgical planning.

Conflict of interest The authors declare that there is no actual or potential conflict of interest in relation to this article.

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