Oromotor disorders in a paediatric neurology unit. Their classification and clinical course

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OROMOTOR DISORDERS IN A PAEDIATRIC NEUROLOGY UNIT. THEIR CLASSIFICATION AND CLINICAL COURSE

Summary. Introduction. *The term 'oro-motor disorders' refers to a group of diseases that predominantly affect sensory inputs, motor systems and movement organization involved in sucking, chewing, swallowing, speech articulation and facial non-verbal communication. Loss of any of the aforementioned functions results in poor social integration and significant quality of life reduction.* Patients and methods. *Retrospective, observational study of 64 patients with oro-motor disorders diagnosed and followed-up at the Child Neurology Service of Vall d'Hebron University Hospital. The oro-motor disorder cause, age at the beginning of symptoms, type of feeding difficulties, type of speech disorders and other associated clinical manifestations were investigated in all patients. Changes in clinical manifestations throughout the period of follow-up in this cohort were analyzed as well.* Results. *Classification of oro-motor disorders in childhood can be achieved combining the etiology and the anatomical location of the underlying disease. Four main groups can be distinguished: due to dysmorphological syndromes; secondary to bilateral perisylvian cortical dysplasias; due to brainstem dysgenesis, and secondary to congenital muscular diseases.* Conclusions. *Establishing the origin, nervous system location and pathophysiology of diseases leading to oro-motor disorders provides clues to natural history and permits anticipation in terms of treatment and care provision. [REV NEUROL 2008; 47: 509-16]*

Key words. *Brainstem dysgenesis. Cortical displasias. Dysmorphological síndromes. Neuro-motor diseases. Oro-motor disorders.*

INTRODUCTION

We seldom realise that such important functions as feeding, speaking or communicating by means of facial expressions are dependent upon the coordinated interaction of a number of structures which several authors call the 'oro-motor system' [1]. Direct control of the oro-motor system takes place in the brainstem. Sensory inputs and motor actions are mediated through neuronal groups (sensory and motor nuclei) located along the brainstem axis. These nuclei, in turn, are under the control of cortical and sub-cortical structures. This sophisticated system can thus be disrupted in many ways and at different levels to give rise to a multitude of disorders, many of which present in childhood.

The term 'oro-motor disorders' (OMD) refers to a group of diseases that predominantly affect the sensory inputs, motor systems and movement organisation involved in sucking, chewing, swallowing, speech articulation and non-verbal facial communication. The adverb 'predominantly' was included in the definition to exclude all congenital or acquired diseases in which OMD was only a minor or marginal clinical manifestation of the patient's disease [2]. On the other hand, even though the definition allows for the inclusion of acquired diseases, the number of such instances in the paediatric age group is so small that, for practical purposes, they are not taken into consideration [3,4].

Not many children have OMD. However, as a group, they deserve special consideration since management of diseases leading to OMD is complex owing to their protracted and varying course, the diverse areas of health care involved and the significant number of resources their treatment requires. Feeding problems, abnormal swallowing, gastro-oesophageal reflux, and pulmonary aspiration of food which may lead to a poor nutritional state are common initial clinical findings in this group of patients, whereas drooling, dysarthria, learning disability and poor social integration are their main late clinical problems, all of which result in poor social integration and a significant reduction in quality of life [2,4,5].

Many aspects of OMD have been reviewed in the literature under different headings: congenital flaccid bulbar paresis [6], dysphagia in childhood [7], congenital suprabulbar and pseudobulbar paresis [8] and feeding or swallowing disorders in children, which are the most commonly found titles in recent paediatric textbooks [9]. Surprisingly, to our knowledge, there are no previous reports in the literature dealing with these disorders from a neurological perspective even though, in most instances, they are caused by nervous system diseases. Establishing the origin, nervous system location and pathophysiology of diseases leading to OMD is no minor issue, since it provides clues to the natural history and allows for anticipation in terms of treatment and care provision.

Following the approach proposed by Illingworth in 1969 [7], over the last 20 years, we have been developing a classification for OMD based on both the aetiology and the anatomical location of the underlying disease (Table I) [9]. The purpose of this paper is to present a review of 64 patients with OMD who were diagnosed and followed up at the Child Neurology Service of the Vall d'Hebron University Hospital and to verify the usefulness of this classification in terms of diagnosis and anticipation of care provision.

PATIENTS AND METHODS

Between June 1985 and June 2007 160 patients were introduced into the database of the Vall d'Hebron University Hospital's Child Neurology Ser-

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vice with the following diagnosis entries: multiple cranial nerve involvement, swallowing disorder, feeding difficulties and speech disorder. Ninety-six of them were excluded from the present study either because of a lack of OMD diagnostic criteria, a shortage of clinical information or insufficient follow-up data. Among these 96 individuals there was a subgroup of patients with perinatal/dysmaturative pathology who, although they fulfilled the OMD diagnostic criteria, could not be included in this study because it was not possible to unambiguously establish the origin or location of their disorder. Revision of clinical charts of all selected patients was carried out and special attention was paid to the following aspects: cause of the OMD, age at the onset of symptoms, type of feeding difficulties, type of speech disorders, other associated clinical manifestations (muscle tone, pyramidal signs, mental status) and changes in their clinical manifestations throughout the follow-up period.

RESULTS

Of all the patients that received attention in the outpatient clinic and were recorded in the database of our unit, 1% of the total number had one of the following diagnoses: multiple cranial nerve involvement, swallowing disorder, feeding difficulties and speech disorder. Half of them fulfilled the diagnostic criteria of OMD. The gender of the 64 patients included in this study was equally distributed among males and females. The follow-up period ranged from 21 days to 23 years (mean: 73.2 months).

A detailed analysis of the clinical information allowed us to classify this group of patients with OMD and divide them into four main groups (Table I). The first group comprised those patients with clinically recognisable dysmorphological syndromes involving the anatomical structures of the oro-motor system. The second group included patients with characteristic signs and symptoms of pseudobulbar involvement: poor or absent voluntary motor control of the lips, tongue, soft palate, pharynx and larynx, dysarthria, dysphagia, drooling and a nasal tone of voice, which frequently associate with cognitive impairment and epilepsy. In all cases, unilateral or bilateral cortical dysplasia was documented by means of MRI studies. The third group consisted of patients with congenital hypotonia and involvement of multiple cranial nerves who, in addition, may present temporomandibular ankylosis, oculo-motor apraxia and mild signs of pyramidal tract involvement. In some of them, brainstem involvement can be demonstrated with neuroradiological studies, while, in the others, lesion localisation is inferred through the clinical manifestations and the results of neurophysiological studies (brainstem evoked potentials, blink reflex and sleep EEG recordings). The fourth group comprised patients with neuromuscular disorders in which the oro-motor involvement was the predominant clinical manifestation. This last group was made up of patients suffering from congenital myotonic dystrophy and congenital myopathies. In the miscellanea group, we included patients with isolated anatomical oro-facial malformations, ex-premature infants or infants with delayed maturation in whom the underlying cause or localisation of the lesion could not be unambiguously ascertained and, finally, all those patients who underwent prolonged mechanical ventilation and/or tracheotomy.

The most relevant clinical and laboratory data of patients included in each of the aforementioned groups is depicted in Tables II, III, IV and V.

Table I. Oro-motor disorders in childhood: classification

Malformative syndromes
Dysmorphologic syndromes
Deletion 22q11.2
CHARGE association (*coloboma, heart defect, atresia, retarded growth, genitourinary problems, ear abnormalities*)
Other (Opitz G-BBB, Goldenhart, Carey-Zineman-Fiter)
Non-syndromic malformative phenotypes
Cerebral hemispherical involvement
Cortical
Sub-cortical white matter
Mixed cortical-sub-cortical involvement
Brain stem involvement
Dysgenetic
Gene mutations
Acquired in-utero
Dysmature
Motor unit disorders
Congenital myotonic dystrophy
Congenital myopathies
Congenital myasthenia syndromes
Other
Miscellaneous
Preterm infants
Isolated anatomical malformations (ie: cleft palate)
Prolonged mechanical ventilation and/or tracheotomy
Abnormal sensory inputs/dysmature

DISCUSSION

Over the last 20 years we have developed a classification for OMD based on both the aetiology and the anatomical location of the underlying disease (Table I) [9]). As can be seen in table II, OMD is a predominant clinical problem in some well recognised dysmorphological syndromes such as velocardiofacial, CHARGE (*coloboma, heart defect, atresia, retarded growth, genitourinary problems, ear abnormalities*), Goldenhart and others. Velocardiofacial syndrome is by far the most common and the one that most commonly exhibits OMD. Hemizygous mutations on chromosome 22 in the q11.2 region are found in 80 to 100% of the reported patients. However, distinction from other syndromes is difficult given that this genetic anomaly has also been described in DiGeorge disease, in CHARGE association and in Cayler's syndrome, among others [10,11]. Velocardiofacial syndrome is characterised by the association of velopalatine dysfunction, cardiac anomalies, a dysmorphic face and psychomotor retardation. The mutation appears 'de novo' in the majority of cases, although in some patients it has been reported as an autosomal dominant trait. In most instances diagnosis is suspected during the work-up investigation of congenital heart diseases. In a study on 251 patients with cardiac defects, 18% had the 22q11.2 deletion [12]. Sucking or swallowing problems are infrequent in early infancy, although regurgitation and respiratory complications are common (5 of our 12 patients). In other instances diagnosis is reached during the work-up study for language disorders or learning disabilities. In a series of 305 patients with mutations in the 22q11.2 and after a prolonged follow-up period, 69% showed structural or functional palatal anomalies which, in all cases, were associated to a nasal tone of voice, and in 77% of patients above five years of age, dysarthria [13]. Despite the fact that the majority of these

Table II. Patients with dysmorphologic syndromes.

No./Sex	Syndrome	Age at diagnosis	Neonatal-Infant		Current age	Childhood-Adolescence			
			Feeding	Other manifestations		Chewing/ Swallowing	Language	Cognitive	Other
1/F	Velocardiofacial	NN	Poor feeding	ASD + VSD, palatal cleft	4 y 3 m	N/N	Dysarthria	ALT	Chronic otitis
2/M	Velocardiofacial	NN	N	Fallot tetralogy	8 y	N/N	Dysarthria	ALT	
3/M	Velocardiofacial	3 y	Poor feeding		3 y 2 m	ALT/N	No	ALT	Otitis media
4/F	Velocardiofacial	10 m	Poor feeding	VSD, resp. infections, GER	6 y	ALT/ALT	Dysarthria	ALT	Hypomimia
5/F	Velocardiofacial	18 m	N	Fallot tetralogy	18 m	–/ALT			Palatal cleft
6/M	Velocardiofacial	22 m			22 m	N/ALT			Hypomimia, drooling
7/F	Velocardiofacial	NN	NGT (15 m)	Fallot tetralogy, resp. infections, GER	9 m				
8/M	Velocardiofacial	4 y 6 m	N	Resp. infections, GER	6 y 10 m	ALT/N	Dysarthria	N	Regurgitations, hypomimia
9/M	Velocardiofacial	NN	N	ASD + VSD, hypotonia, choking, GER	21 m	N/N		ALT	
10/M	Velocardiofacial	3 y 10 m	N	Resp. infections	9 y	N/N	Dysarthria	ALT	Velopalatal insufficiency, nasal speech, drooling, right VI cranial nerve palsy, frontal bilateral dysplasia
11/M	Velocardiofacial	3 y	N (slowly)	Hypotonia, resp. infections	3 y 6 m	N/ALT	Dysarthria	N	
12/M	CHARGE	NN	NGT (4-18 m)	Hypotonia	8 y 6 m	ALT/N	N	ALT	Drooling, SNHL
13/F	Opitz-Frias	1 m	NGT → gastrost. (5 a)	Phenotype anomalies, resp. infections, apneas	5 y	ALT/ALT	N		Microcephaly, microretrognathia
14/M	Ochoa	2.5 m	NGT	Resp. infections, GER	10 y 9 m	N/ALT	N	N	Velopalatal insufficiency, hydronephrosis
15/F	BPES	NN	N	Polimalformative	7 y	ALT/N	No	ALT	Hypomimia, velopalatal insufficiency, TMA
16/F	del(2)(q32.2-q35)	NN	NGT	Microcephaly, choking, palatal cleft	4 m				

ALT: altered; ASD: auricular septal defect; BPES: blefarofimosis-ptosis-epicantus inversus syndrome; CHARGE: coloboma, heart defect, atresia, retarded growth, genitourinary problems, ear abnormalities; F: female; gastrost.: gastrostomy; GER: gastro-esophageal reflux; M: male; m: months; N: normal; NGT: naso-gastric tube feeding; NN: neonatal; resp.: respiratory; SNHL: sensoryneural hearing loss; TMA: temporo-mandibular amkilosis; VSD: ventricular septal defect; y: years.

patients suffer from mild to moderate mental retardation, language is disproportionately disturbed in relation to the rest of their abilities. Moreover, the propensity of these patients to suffer chronic serous otitis contributes to their poor speech performance. It has also been reported that these patients, during infancy, reject solid feeding or experimenting with different food textures. Feeding and speech problems have been attributed to velo-palatine insufficiency, as well as their lack of coordination and/or hypercontractility of the pharyngeal and oesophageal segments [15].

In 1954, Worster-Drought first described a group of individuals with a congenital pseudobulbar syndrome characterised by oro-facial paresis, dysarthria, difficulties in voluntary motor control of the tongue and, characteristically, dissociation between cortical and sub-cortical motor control [8,16]. in 1993, Kuzniecky et al [17] introduced the term ‘congenital bilateral perisylvian syndrome’ (CBPS) to refer to a group of patients with pseudobulbar signs and symptoms associated with bilateral perisylvian polymicrogyria demonstrated by cerebral MRI. Patients described by Kuzniecky differed from those reported

Table III. Patients with bilateral cortical dysplasia.

No./Sex	PMG distribution	Age at diagnosis	Neonatal-Infant		Current age	Childhood-Adolescence					
			Feeding	Muscular tone		Chewing/ Swallowing	Language	Hypotonia/ Pyramidal signs	Cognitive	Epilepsy	Other
1/M	Frontal	4 m	N	Hypertonia	2 y 6 m (†)	N/ALT	Absent	Pyramidal signs	ALT	Clonic seizures, extensor spasms	Clonazepam (pregnancy), sagital cranio-synostosis, poli-malformative syndr.
2/M	Frontal-parietal	2 y	N	N	16 y 10 m	N/N	Dysarthria		ALT	Generalyzed	Drooling
3/M	Left frontal-parietal	2 y	N	N	7 y	N/N	Dysarthria	N	ALT	Focal	Neonatal fits, behaviour disorder, facial diplegia
4/F	Frontal-perisylvian		N	N	12 y	ALT/ALT	Dysarthria	Pyramidal signs	ALT	No	Drooling, microcephaly
5/M	Frontal-perisylvian	NN	N	Hypotonia	4 y 6 m	ALT/ALT	Absent	Hypotonia	ALT	Flexor spasms	10p trisomy-9p monosomy, CNS complex malformation, poli-malformative syndr., drooling, GER
6/F	Perisylvian-temporal	< 1 y	N	N	12 y	ALT/N	Absent	Pyramidal signs	ALT	No	CMV infection (pregnancy), resp. infections, drooling
7/M	Perisylvian	6 m	N	N	5 y	N/N	Absent	Pyramidal signs	ALT	Extensor spasms	Facial diplegia, drooling
8/M	Posterior perisylvian	NN	N	N	8 y	ALT/N	Dysarthria	Pyramidal signs	ALT	No	CNS complex malformation, poli-dactily, stereotypies
9/M	Posterior perisylvian	2 y 8 m	N	N	7 y	ALT/N	Dysarthria		ALT	No	Congenital hypo-tiroidism, NF1, facial diplegia, drooling
10/M	Diffuse	8 m	N	Hypotonia	12 y	N/N	Dysarthria	Pyramidal signs	ALT	No	Drooling, cianosis episode, macrocephaly
11/F	Perisylvian + pachygyria	18 m	N	Hypotonia	6 y	ALT/N	Absent	Pyramidal signs	ALT	No	Drooling, facial diplegia, phenotype anomalies
12/F	Perisylvian + pachygyria	2.5 y	N	N	8 y	N/N	Dysarthria	Pyramidal signs	ALT	No	Resp. infections, drooling, facial diplegia

ALT: altered; CMV: citomegalovirus; CNS: central nervous system; F: female; GER: gastro-esophageal reflux; M: male; m: months; N: normal; NF1: neurofibromatosis type 1; NN: neonate; PMG: polymicrogiria; resp.: respiratiry; syndr.: syndrome; y: years.

by Worster-Drought in their greater rate of neurological involvement, cognitive impairment and, particularly, epilepsy. In the last few years, investigation of familial cases of bilateral polymicrogyria has led to the association of this finding with mutations at the Xq28, Xq22 (gene *SRPX*) loci, mutations in the *MECP2* gene or micro-deletions at the 22q11.2 loci [18]. Although the origin of bilateral polymicrogyria in most instances cannot be established, genetic factors, prenatal infections and vascular lesions are considered to be the most likely causes of this disorder. Recently, several authors have suggested the possibility that Worster-Drought and CBPS are, in fact, different clinical forms of the same disorder and not two separate clinical entities [5,18,19].

Among our twelve patients with bilateral cortical dysplasia (Table III), clonezapan exposure during the first week of gestation (patient 1), trisomy 10p-monosomy 9p (patient 5) and congenital CMV infection during the second trimester of gestation (patient 6) were found as possible causes of their CNS malformation. Interestingly enough, patient 3, in whom unilateral fronto-parietal polymicrogyria was only demonstrated in MRI studies, experienced a worsening of his pseudobulbar symptoms during a period of poor control of his focal epilepsy [21]. Clinical manifestations in this group of patients appeared during the first two years of life (11/13 patients) and, surprisingly, global developmental delay or abnormal muscle tone were the most commonly recorded complaints. No significant feeding

Tabla IV. Patients with brainstem dysgenesis.

No./Sex	Cranial nerves involved	Age at diagn.	Neonatal-Infant		Current age	Childhood-Adolescence				
			Feeding	Muscular tone		Chewing/ Swallowing	Language	Hypotonia/ Pyramidal sings	Cognitive	Other
1/M	III, IV, V, VI, VII, VIII	NN	Poor feeding	Hypotonia, TMA	21 d (†)					Non-spontaneous, respiration, clubfeet
2/F	V, IX, X	NN	NGT (8 m) → gastrostomy	Hypotonia, TMA	5 y	ALT/ALT	No	Hypotonia	ALT	Oculomotor apraxia, microretrognathia, resp. infections
3/M	VII (R > L), IX, X	2.5 m	Gastrostomy (2.5 m)	Hypotonia, TMA	6 y	ALT/ALT	No	Pyramidal signs	ALT	Oculomotor apraxia, GER, resp. infections
4/F	V, VII, IX, X	NN	NGT (21 m)	Hypotonia, TMA	23 y	ALT/N	Dysarthria	Pyramidal signs	N	Oculomotor apraxia, microretrognathia
5/F	V, VII, IX, X	NN	NGT (4-12 m)	Hypotonia, TMA	16 y	ALT/ALT	Dysarthria	Pyramidal signs	ALT	Oculomotor apraxia, facial diplegia, nasal speech, drooling
6/M	V, VII, IX	NN	NGT (10 m)	Hypotonia → pyramidal signs, TMA	15 y	ALT/N	Dysarthria	Pyramidal signs	ALT	Oculomotor apraxia, microretrognathia, kyphoscoliosis
7/F	III, IV, V, VI, VII, VIII	NN	NGT (3 y 6 m)	TMA	2 y 6 m (†)	NGT			N	Oculomotor apraxia, GER, resp. infections
8/M	III, VI, VII, IX,X	NN	Poor feeding	Hypotonia	4 m (†)					Facial diplegia, epileptic seizures (cortical dysplasia)
9/F	VI, VII, IX, X, XII (R > L)	NN	NGT (2-8 m)	Hypotonia	7 y	ALT/ALT	Dysarthria	Hypotonia	N	Clubfeet, resp. infections
10/M	VI, VII (L > R), IX, X, XII	NN	NGT (d), poor feeding	Hypotonia, TMA	6 m	–/ALT				GER, microretrognathia, microglossia, dysphonic cry
11/M	VI, VII, IX, X, XII	NN	NGT	Hypotonia, TMA	1 m	–/ALT		N		Facial diplegia, microretrognathia
12/F	VII (R), IX, X, XII	NN	NGT (1.5 m)	Hypotonia	27 m	ALT/ALT	No	Hypotonia	ALT	Left hemiparesis
13/F	VII, IX, X	NN	Gastrostomy (6 m)	Hypotonia → pyramidal signs	9 m	–/ALT				Facial diplegia, resp. infections
14/F	VI, VII bilateral, XII R	NN	NGT (2 m)	N	13 y	ALT/ALT	Dysarthria	N	ALT	Clubfeet, microretrognathia, microstomy, scoliosis
15/M	VI, VII bilateral, VIII R	NN	N	N	23 m	N/N	Dysarthria	N	N	GER, clubfeet
16/M	VI, VII	2 y	N	N	3 y 6 m	N/N	Dysarthria	N	N	Nasal speech
17/M	VII (L > R)	18 m	Poor feeding	Hypotonia	4 y	ALT/N	Dysarthria	N	N	Facial diplegia. drooling, jaw reflex ++
18/F	IX, X	NN	NGT (2.5 y) → gastrostomy	Hypotonia	5 y	Gastrostomy (2.5 y)	N	N	N	Microretrognathia, drooling, nasal speech

ALT: altered; d: days; F: female; GER: gastro-esophageal reflux; L: left; M: male; m: months; N: normal; NGT: nasogastric tube feeding; NN: neonatal; R: right; TMA: temporo-mandibular ankylosis; y: years.

problems were reported in any of our patients during the first years of life. These findings are in disagreement with those described by Kuzniecky et al [16] or by Clark et al [5], who reported feeding difficulties during the neonatal period in 64.5% and 60% of their patients, respectively.

Chewing and swallowing problems most commonly associate with perisylvian polymicrogyria, although, according to our data, precise clinical neuroradiological correlations cannot be drawn in patients with bilateral cortical dysplasia. Cognitive functions were involved in all our patients, the degree of cognitive impairment being directly correlated, as Worster-Drought already pointed out, with the extent of speech and language disturbances. In some of these patients, facial hypomimia with diminished facial expression plays an important role in their social interactions, as they are perceived as severely retarded individuals. Five out of twelve (41.6%) of our patients experienced

Tabla V. Patients with neuromuscular diseases.

No./Sex	Disease	Age at diagnosis	Neonatal-Infant		Current age	Childhood-Adolescence				
			Feeding	Muscular tone		Chewing/ Swallowing	Language	Hypotonia	Cognitive	Other
1/F	Myotonic dystrophy	NN	NGT 10 d	Hypotonia				No		Microretrognathia, polypnea
2/M	Myotonic dystrophy	NN	Poor feeding	Hypotonia	15 y	N/N	N	Yes	ALT	Hypomimia, nasal speech
3/F	Myotonic dystrophy	5 y			9 y	N/N	Dysarthria	No	N	Hypomimia
4/M	Myotonic dystrophy	NN	N	Hypotonia	7 y 6 m	N/N	Dysarthria	Yes	ALT	Hypomimia, drooling, clubfoot, cianosis episode
5/F	Myotonic dystrophy	NN	Poor feeding	Hypotonia	7 y	N/N	Dysarthria	No	ALT	Bilateral femur fracture, clubfoot, facial diplegia, nasal speech
6/F	Myotonic dystrophy	NN	N	Hypotonia	14 y	N/N	Dysarthria	Yes	ALT	Hypomimia, nasal speech
7/M	Myotonic dystrophy	NN	Poor feeding	Hypotonia	9 y	N/N	NO	No	ALT	Nasal speech
8/M	Myotonic dystrophy	NN	N	Hypotonia	3 y 6 m	N/N	Dysarthria	No	ALT	Nasal speech
9/M	Myotonic dystrophy	NN	Poor feeding	Hypotonia	4 y 3 m	N/N	Dysarthria	No		Hypomimia
10/F	Myotonic dystrophy	NN	N	Hypotonia	9 y	ALT/N	N	No	ALT	Nasal speech, hypomimia
11/F	Myotonic dystrophy	NN	NGT (1,5m)	Hypotonia	12 y	N/N	N	No	ALT	Nasal speech
12/F	Spinal musc. atrophy	NN	NGT (6m)	Hypotonia	14 m	N/N		Yes	ALT	
13/M	Spinal musc. atrophy	NN	NGT (6,5m)	Hypotonia	9 y 6 m			No		GER, clubfoot, hypomimia
14/F	Nemaline myopathy	NN	NGT (36m)	Hypotonia	19 y	ALT/N	Dysarthria	Yes	N	Nasal speech, otitis media
15/ F	Nemaline myopathy	NN	NGT (7m)	Hypotonia	5 y 6 m	ALT/ALT	Dysarthria	Yes	N	GER, drooling
16/F	FSHD	NN	N	Hypotonia	13 y	ALT/ALT	N	Yes	N	Hypomimia
17/F	Idiopathic myopathy	4 y	N	N	10 y 4 m	ALT/ALT	Dysarthria	Yes	N	Drooling, distal hyperlaxity
18/F	Congenital myastenic syndrome	NN	NGT 18 d	Hypotonia	> 18 y	N/N	N	No	N	Cubfoot, tachypnea

ALT: altered; d: days; F: female; FSHD: facio-scapulo-humeral dystrophy; GER: gastro-esophageal reflux; M: male; m: months; musc.: muscular; N: normal; NGT: nasogastric tube feeding; NN: neonatal; y: years.

epileptic seizures, and three of them had infantile spasms during the first year of life. Kuzniecky et al reported epilepsy in 85% of their patients and in sixteen out of twenty-seven patients this was resistant to medical treatment. Other clinical manifestations found in our group of patients were drooling in 9 out of 12 cases, facial diplegia in 5 patients, mild signs of pyramidal tract involvement in 8 out of 12, complex CNS malformations in 2 and non-recognisable polymalformative syndromes in 2 other patients.

Over the last 23 years we have identified 18 patients (Table IV) in whom the lesion giving rise to the OMD is located in the brainstem. In order to designate all these patients, several years ago [21] we proposed the term 'brainstem dysgenesis', based on the location of their lesion and the notion that a common physiopathological mechanism (dysgenesis) was behind each of the different clinical presentations. In the majority of cases, the causes of the dysgenesis are prenatal destructive accidents, of which those of a vascular nature are the most frequent. There is no doubt, however, that in some brainstem dysgenesis patients the clinical manifestations have a genetic origin, as is the case of the Atabascan brainstem dysgenesis and 'Bosley-Salih-Alorainy syndrome' [22], which have been associated to mutations of the *HOXA1* gene or the reported familial cases of Möbius syndrome. Neonates with transient clinical findings similar to those of brainstem dysgenesis have been reported [23] and they are considered to suffer short-lived dysfunctions due to retarded maturation of the oro-motor system structures located in the brainstem.

According to the extent of brainstem involvement, several clinical forms can be distinguished:

– *Lethal forms (patient 1):* the most severe, associated with respiratory failure and death occurring in the first month of life.

– *Diffuse forms (patients 2-13):* includes those patients with hypotonia and involvement of multiple cranial nerves, mild pyramidal signs, temporomandibular ankylosis and feeding problems. Five of these patients, who also showed transient oculo-motor apraxia, have already been reported [24].
– *Intermediate forms (patients 14, 15 and 16):* match the classical description of Möbius or Pierre-Robin syndromes.
– *Restricted forms (patients 17 and 18):* with isolated bilateral involvement of one of the cranial nerves, which reflects the disrupted segment of the brainstem.

Finally, for each brainstem dysgenesis form there are asymmetrical variants (patients 10, 12, and 17), for whom the disruptive vascular cause is the most likely origin.

Poor sucking, swallowing and feeding problems were present soon after birth in the majority (16 out of 18) of our patients. Nasogastric tube feeding was required in 10 and a gastrostomy had to be carried out in 3 of them. Muscle hypotonia, predominantly axial in distribution, had also been present since birth in most patients (16 out of 18). Remarkably, this decrease in muscle tone is often associated with signs of pyramidal tract involvement (increased DTR, Achilles clonus). In some patients, there is poor handling of feeding and secretions with episodes of shock and apnoea since birth.

Muscle tone and motor abilities of patients with brainstem dysgenesis progressively improve with passing years and they are left with feeding, chewing, dysarthria and non-verbal facial communication problems. Half the patients who could reliably perform an IQ test gave abnormal results, which is an interesting observation that reinforces the issue of the brainstem participating in high cortical functioning. Alternatively, the low IQ of these patients could be explained by a mixed involvement of lower motor neurons, brainstem and cortical structures, as has been suggested in recent publications [25].

There are few reports on oro-motor dysfunction and neuromuscular disorders; this probably reflects the fact that the focus is placed on the intimate origin of the underlying disease rather than on this particular association of clinical signs. Nevertheless, decreased muscle tone, sucking, swallowing and respiratory difficulties are present since birth in many congenital neuromuscular diseases. In our series of patients 11 out of 18 had these symptoms and, of those, 7 required nasogastric tube feeding for varying periods of time. The severity of the clinical manifestations is closely related with the type of underlying neuromuscular disorder. Hypotonia, sucking, swallowing and respiratory difficulties are quite prominent since birth in the fatal forms of some congenital myopathies or after several months of life in type I SMA, but it can be milder in other muscle diseases such as congenital muscular dystrophy or myotonic dystrophy. As a general rule, muscle tone and strength improve in the surviving patients and, following this amelioration, the respiratory and feeding problems of these patients also improve. As patients get older, dysarthria secondary to the anatomical changes in the oral cavity as well as velo-palatine insufficiency are the predominant clinical signs. In congenital myotonic dystrophy, the most common disease in this OMD group, muscle hypotonia, facial diplegia and feeding problems can be severe during the neonatal period, particularly if this disorder is associated with prematurity. After infancy, patients can be recognised by the nasal tone of voice, diminished facial expression and their poor school performance [26].

Early diagnosis and classification of OMD is crucial, since it allows genetic counselling for future pregnancies in polymalformative syndromes and other genetically determined diseases as well as anticipation in the diagnosis of other associated clinical manifestations, which in turn enables appropriate preventive treatments to be implemented. In this regard, it is worth mentioning that most patients with moderate to severe OMD show a poor response to standard speech therapy, particularly if there are associated cognitive deficits, and therefore alternative methods of communication must be taken into consideration early on in the process. This approach may prevent loss of self-esteem and a better quality and quantity of communication from the early stages of life.

Finally, clinical management of all these patients must be multidisciplinary owing to the involvement of many different systems or functions (feeding, speech, motor). On the other hand, all these disorders show changing aspects with the passing of time and, hence, social care and requirements for resources need to be taken into consideration throughout the whole life span of these individuals.

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TRASTORNOS OROMOTORES EN UNA UNIDAD DE NEUROLOGÍA PEDIÁTRICA. CLASIFICACIÓN Y EVOLUCIÓN CLÍNICA

Resumen. Introducción. *El concepto 'trastornos oromotores' engloba al conjunto de enfermedades que afectan de forma predominante a las estructuras que intervienen en la movilidad facial y orofaríngea, y que son imprescindibles para una correcta mecánica de la alimentación y para la articulación del lenguaje y la expresividad facial. La alteración de estas funciones puede afectar significativamente a la calidad de vida y a la integración social de estos pacientes.* Pacientes y métodos. *Estudio descriptivo retrospectivo de 64 pacientes diagnosticados de trastornos oromotores, controlados en el Servicio de Neurología Pediátrica del Hospital Universitari Vall d'Hebron. De cada uno de ellos, detallamos: etiología, edad de presentación, información acerca del trastorno de la alimentación, de la articulación del lenguaje y de posibles manifestaciones asociadas, y evolución durante el período de seguimiento de los diferentes aspectos clínicos.* Resultados. *La revisión de la casuística del servicio nos ha permitido clasificar a los enfermos afectos de trastornos oromotores en función de la localización de la lesión y su enfermedad de base, distribuyéndolos en cuatro grupos principales: pacientes con síndromes polimalformativos, con afectación cortical perisilviana, con disgenesia troncoencefálica y con afectación del sistema nervioso periférico.* Conclusiones. *La clasificación de los pacientes afectos de trastornos oromotores y el estudio de la evolución natural de cada uno de los grupos facilitan el abordaje y permiten optimizar el manejo y realizar una prevención adecuada de las posibles complicaciones de este tipo de pacientes. [REV NEUROL 2008; 47: 509-16]*

Palabras clave. *Afectación cortical. Disgenesia troncoencefálica. Enfermedades de neurona motora. Síndromes polimalformativos. Trastornos oromotores.*