SWALLOWING AND FEEDING IN CHARGE SYNDROME: FOR THE PHYSICIAN

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ANOMALIES SPECIFIC TO CHARGE

Swallowing difficulties, one of the major features of CHARGE syndrome, particularly in the first years of life, occurred in 96% of our children with CHARGE[1]. Other authors report between 31and 88%.[2,3] The main causes are as follows:

Poor suck-swallow coordination. Neonatal Brain Stem Dysfunction (NBSD):

Neonatal brain stem dysfunction is the major cause of feeding problems in CHARGE. NBSC, previously reported in children with Pierre Robin sequence (4-6), is a group of four types of symptoms involving the supranuclear region of the IXth, Xth and XIIth cranial nerves. These symptoms, important in the first two years of life, seem to be related to a developmental defect of the suck-swallow central pattern generator in the solitary tract.[7]

(i) sucking and swallowing skills disorder,
(ii) esophageal dyskinesia,

Clinically, both of these symptoms result in poor suck, prolonged feeding time, milk aspiration, unexplained cries during bottle feeding, nasal reflux, regurgitation and ALTE (Apparent Life Threatening Event) during feeding. They may lead to pharyngeal congestion, aspiration pneumonia and failure to thrive. The esophageal dyskinesia is responsible for gastroesophageal reflux (GER) that is poorly managed by classical medical treatments.

(iii) glosso-pharyngeo-laryngomalacia
(iv) sympathetic-parasympathetic cardiac rhythm dysregulation.

The glosso-pharyngeo-laryngeomalacia is responsible for obstructive apneas or hypopneas and subconsequent hematosis while the vagal dysregulation may result in ALTE, induced by all previously noted symptoms. Sucking and swallowing incoordination of NBSD resolves when corticalization of feeding occur, after 6 mo. of life.

Facial palsy

The facial nerve plays a role in face and lip movements which increase sucking problems. Facial palsy in CHARGE is almost always unilateral and peripheral due to dysgenesis of the VIth nerve extra bulbar pathway. Determining the origin of facial palsy is important for prognosis and electromyogram may help. Facial palsy may show little or no improvement with age and may be responsible for further aesthetic problems.

Malformations of esophagus, larynx or choanae ; cleft lip and palate.

Anatomical malformations of the organs involved in sucking, swallowing and breathing are also responsible for swallowing and are discussed in other sections of this manual.

Hyposmia

Abnormal olfactory lobes have been noted in CNS autopsy of CHARGE patients[2] and recently, the common embryological origin of olfactory tracts and hypothalamus involved in hormonal defects (LHRH) led to a better MRI analysis of that brain region, showing frequent anomalies.[8,] personal data

Even at birth smell seems to be involved in feeding behavior and increases appetite. This is more evident after corticalization of feeding, i.e. at the end of the first year. The clinical consequences of hyposmia need to be further defined, even in young infants, by perfecting olfactometric scales for children before language acquisition. Olfactory competence in CHARGE has been poorly investigated for both technical and medical reasons: i) olfactometric scales requiring good language cannot be performed by a normal child before 10 years of age and ii) hyposmia is a minor problem that escapes notice.

Exogenous factors

Finally, feeding difficulties may also be secondary to exogenous factors such as cardiac or pulmonary dyspnea, effects of initial nasogastric tube feeding whatever the reasons of the enteral nutrition and deleterious effects of the long initial hospitalization both on the child and on altering the precocious mother-child emotional bonds.

DIAGNOSTIC TESTS

Sucking and swallowing disorders are mainly evaluated by clinical means. A guided anamnesis, a good history and observation of the child during feeding are the best tests. Cineradiography may be dangerous and is not sensitive enough in mild cases. It may help determine when to restart an oral feeding program when aspiration risks diminish. Succimetry may be interesting but, in our hands, remains a research procedure.

Neonatal brain stem dysfunction can be investigated by its peripheral effects.

i) Esophageal dyskinesia may be investigated by an esophageal manometry, which provides specific information on abnormal esophageal motor control: hypertonia or achalasia of the lower esophagus sphincter, abnormal coordination of pharyngeal contraction and the upper esophageal sphincter. These features are not constant (95% of investigated children in our series had at least one abnormal manometric criterion, 60% a specific anomaly and the rest a less specific abnormal motility of the esophagus). Investigations of the neurologically-based GER are only necessary when surgical treatment is considered. X-ray barium transit is useful but pH meter readings are less so since results may be normal even if vomiting is evident.

ii) Laryngeal endoscopy may show specific aspects of hypotonia of the tongue base, pharynx walls and larynx. It may also show salivary stasis and peptic inflammation of the larynx and trachea secondary to gastric acid reflux and feeding aspirations.

iii) A 24-hour electrocardiogram recording (Holter) with ocular compression test may help to evaluate a vagal hyperactivity.

iv) Evoked potentials of the brain stem may show abnormal delay in tracing, particularly during the first steps of the auditory evoked potentials (AEP) used in

assessing hearing loss. Swallowing disorders by themselves do not require this investigation in practical terms.

v) Facial and endobuccal electromyography (EMG) as well as dynamic EMG (when it is not dangerous) during bottle feeding may be important to determine the origin of cranial nerves defects. Most often, recording of the 7th cranial nerve shows peripheral palsy. Recordings of 9th 10th and 12th nerves separately are normal but dynamic EMG during bottle feeding shows poor coordination between muscles innervated by the 9th and 10th nerves respectively.[9] This test mainly has theoretical interest but it is quite aggressive and needs a specialized operator.

vi) Finally, investigations of olfactory abilities may be performed with adapted scales (personal data) or by MRI of the olfactory lobes. Again, these investigations are not required for proper treatment of swallowing difficulties.

MEDICAL CONSEQUENCES

Medical consequences of swallowing disorders are numerous. They alter pulmonary status, induce chronic bronchitis with a risk of hypoxemia and hypercapnia and adversely affect cardio-pulmonary vascularization, quality of sleeping and psychomotor development. Feeding aspirations and stasis in the pharynx worsen respiratory obstructive syndrome. Discomfort and pain induced by swallowing disorders lead to a decreased intake and failure to thrive. Sucking and swallowing disorders alter mother-child relations and increase the risk of further anorexia even when organic problems are solved.

Personal data. 30 children with CHARGE from 1 to 9 years

- Adapted diet and feeding procedures only 3 children
- Nasogastric tube only 7 children (3 precocious deaths)
- Gastrostomy and GER surgical treatment 20 children
- Mean age of artificial nutrition weaning 3 years (min 16 mo, max 7 yr)
- Mean age of normal eating recovery (if reached) 5 years
- Number who do not eat after 6 years of age 2 children

MEDICAL AND NON MEDICAL MANAGEMENT *primum non nocere*

Apart from surgical treatment of esophagus and upper airway anatomic malformations, swallowing disorders have no radical medical therapy. Spontaneous resolution of functional disorders of sucking and swallowing is generally good and the major challenge is to wait for their natural improvement in the best conditions. One should recognize feeding disorders early in order to prevent deleterious consequences, reduce all contributing factors, teach feeding by multisensory and physiotherapeutic means and support parents' psychologically:

- Ask the mother for sucking skills disorders, even in the neonatal period:

- Improve sucking and swallowing mechanic, when it is not too poor, by using soft and largely perforated nipples, thickened milk, small meals and avoiding forced feeding.

- If swallowing disorders have respiratory or growth consequences, enteral nutrition is required, either exclusively if aspirations are present or as a night supplement when possible. Enteral nutrition avoids the respiratory consequences of feeding aspiration and induces proper weight gain.

- The choice between nasogastric (NG) tube and gastrostomy depends on the age of the child and the severity of the symptoms. Initially, NG tube feeding is started but, after few months or earlier if the procedure is not well tolerated. If the troubles persist, NG tube should be changed to a gastrostomy combined with anti-reflux surgery.

- Good airway clearance is essential to allow swallowing. Trachoeostomy is often necessary and does not alter swallowing. On the contrary, it provides better ventilation and allows tracheobronchial drainage.

- Non-medical management of feeding teaching is detailed in the parents' section.

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SWALLOWING AND FEEDING IN CHARGE SYNDROME: PARENT INFORMATION

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NORMAL STRUCTURE AND FUNCTION

Feeding is both one of the more essential and the more complex of the mammalian functions. Feeding of children can be divided into two periods :

1. The sucking and swallowing reflex period lasts from fetal life to the age of 6 months and essentially requires the good organization of sensory and motor pathways going and coming from the face and the mouth to the brain stem.

2. The corticalized feeding period, lasting from the middle of the first year to adulthood, involves voluntary processes for the feeding orally and are influenced by sensory, psychological and environmental factors. Adult feeding processes are matured between 2 and 3 years of age.

Moreover, appetite and caloric intake are regulated by several neural mediators coming from the intestinal tract and the CNS (hypothalamus, thalamus, cortex). For children with CHARGE syndrome, the main problems seem to occur during the first period, during the brain stem development and organization. We do not know specific treatment for these disorders so the aim is to wait for the second period when feeding improves on its own.

TESTS USED AND SPECIALISTS

Swallowing disorders are generally very well evaluated by the mother. From their child's first days of life, mothers may observe poor sucking, inability for the baby to be breast fed, discomfort with bottle, unexplained cries, bottle refusal or prolonged feeding time, skin color changes during feeding, breathing difficulties or cough increased by feeding, frequent spitting up, nasal reflux or poor weight gain.

The only two obligatory investigations are, in my opinion, a chest x-ray to detect pneumonia aspirations and an examination of the airway by a competent pediatric otorhinolaryngologist (ENT).

Other investigations may be useful to characterize the origin, the mechanism and the severity of the symptoms. They are performed differently depending on the child and the medical team (see doctor section).

MANAGEMENT

Swallowing difficulties are as badly tolerated by the child as by his/her mother. Young infants affected with sucking, swallowing and breathing difficulties are not able to grow, to develop and to explore their environment in a proper way. Moreover, swallowing and feeding disorders cause great parental anxiety, for both medical and psychological reasons.

Parents have an important role regarding swallowing difficulties in pointing out the signs to the doctor. Parents need to understand that what appear to be very drastic and aggressive techniques (nasogastric (NG) tube, gastrostomy, tracheostomy and no oral feedings) would improve their child's comfort. These medical options are not easy to accept, especially when the problems do not seem to be major ones. Performing surgery may make their child seem more gravely affected and give a feeling of failure to the caretaker. The advantages of these interventions are observed after they done. Discussing options with other parents of children with CHARGE or looking at other children who have had these procedures may help the families during these periods.

Parents also have a great role in the non-medical management of these difficulties by learning how to prevent worsening of the feeding problems and learning early how to adapt the teaching of a normal feeding by working with a Feeding Team. First by their positive attitude without guilt that they are at fault, no forced feeding and no excessive anxiety. Second by their presence and involvement in medical care, using adapted bottles, breast exclusion, by handling, massage or any methods to keep close physical contacts with the baby.

Learning to feed by mouth

After several months of poor coordination of sucking and swallowing, children may be frightened by food. The first step consists of avoiding bad experiences with food. Non-nutritive sucking with a pacifier is useful. Then, the child must get acquainted again with his feeding instrument, that is to say his face, lips, gums. These vulnerable areas have to be approached with soft massage, by mouth games, etc. and always with close contact with the child. The pleasure of feeding has to be taught using pleasant and intense smells (even if the child's sense of smell seems to be poor), colors or tastes.

Food with a smooth texture should be given with a soft plastic spoon. Problems tolerating solid food can last several years. From the end of the first year, solid pieces may be offered to the child, allowing himself the peace from his hand to his mouth, in a totally voluntary procedure.

After the period of aspiration risk, drinking must be taught too, playing with pure water in a glass. Chewing must be stimulated too by elastic texture, placed in the lateral part of the mouth.

The management of feeding problems is a subjective procedure, depending on team and culture and requiring multidisciplinary interveners. In our opinion, this management must be a medical concern from the first week of life of a children affected with CHARGE syndrome.