SWALLOWING EVALUATION IN INFANTS WITH CONGENITAL HEART DISEASE AND DOWN SYNDROME : CLINICAL STUDY CASES

Avaliação da deglutição em lactentes com cardiopatia congênita e síndrome de Down: estudo de casos

Deborah Fick Böhm Fraga[1], Karine da Rosa Pereira[1], Sílvia Dornelles[1], Maira Rosenfeld Olchik[1], Deborah Salle Levy[1]

ABSTRACT

This study had the aim to perform clinical swallowing evaluation in infants diagnosed with Down syndrome and congenital heart disease (complete atrioventricular canal) suspected to have swallowing disorders hospitalized in unit 2A and Pediatric ICU of instituition.It is a descriptive and qualitative study in which was possible to assess two infants during the proposed period. It was used a sample profile protocol to collect data about clinical history and diagnoses from patients records and the Instrumento de avaliação para prontidão do prematuro para alimentação oral by Fujinaga (2002) for clinical swallowing evaluation. The pacients presented results of oropharyngeal dysphagia and low scores invalidating exclusively oral feed. The treatment was oral stimulation and oral feeding volume controlled. The findings contribute to the literature regarding the correlation between dysphagia, congenital heart defects, genetic syndromes and myofunctional disorders. It is concluded that dysphagia was presented as a difficulty symptom to safe and effective oral feed for all infants studied. Prospective larger researches are needed to contribute with this clinical cases study and thus identify other risk factors for dysphagia and specific treatment for children with Down syndrome and swallowing disorders associated.

KEYWORDS: Deglutition Disorders; Down Syndrome; Heart Defects, Congenital; Infant; Deglutition; Evaluation

INTRODUCTION

In recent years, great importance has been given to related oropharyngeal dysphagia due to the high prevalence of this symptom in adult and pediatric studies. Dysphagia or swallowing disorder refers to the difficulty in passing the bolus from the oral cavity to the stomach which sometimes makes it difficult or impossible to secure the food ingestion. There are a few published studies and surveys that measure the incidence and prevalence of dysphagia in pediatric patients with congenital heart disease. The swallowing disorders usually present in cases of multiple diagnoses, syndromes and associated comorbidities, suffering conditions influence the overall development of children[1].

Down syndrome is the most frequent chromosomal alterations, described in 1866 by John Langdon Down, as microgenia, macroglossia, epicanthu , oblique palpebral fissures, shorter limbs, a single transverse palmar crease, poor muscle tone, mental retardation and learning difficulties, which came to characterize the trisomy 21. It is checked one in every 800 live births and the risk for occurrence increases significantly in the presence of advanced maternal age [2,3]. In addition to the phenotypic features that include structural changes of the

(1) Universidade Federal do Rio Grande do Sul – UFRGS, Porto Alegre, RS, Brasil.

Work performed at the Institute of Cardiology of Rio Grande do Sul IC / FUC – RS.

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face , infants with Down syndrome, in 43% of cases, has congenital heart disease that is often corrected by surgery[3].

Congenital heart disease refers to abnormalities that occur before birth in the structure or function of the heart, it is estimated that every 1,000 births four to 50 infants possess this amendment, which still represents a major cause of infant mortality in the United States and the second largest in Brazil[4-6]. The necessary surgeries usually weaken the general health and the quality of life of the patients[5,6].

A child with Down syndrome has severe motor and functional involvements in regions related to the swallowing process as a result of the morphological features described above.

Another factor that can be cited is the frequent exposure to corrective surgery for congenital heart defect with its complications (ventilation, hospitalization for a prolonged period, use of feeding tube, etc.). These conditions may be of a important risk to the dysphagia symptom affecting children with this specific syndrome, often can cause nutritional deficiencies, dehydration, sensorimotor impairment and tracheal aspiration, which represents risk to the general health of the patients [1,7,8].

In order to contribute to this field of research, the present study aimed to investigate the swallowing process and possible changes through clinical evaluation in two infants with Down syndrome and congenital heart disease.

CASE PRESENTATION

This is a descriptive-qualitative research approved by the Ethics and Research Institute of Cardiology of Rio Grande do Sul Committee (IC / FUC) under number 4603/11.

The Statement of Informed Consent Form was duly signed by the legal guardian of each individual prior to the performance of speech-language evaluation.

All infants with suspected swallowing difficulty were included, and submitted to the speech therapy service, with diagnoses of congenital heart disease and Down syndrome, hospitalized in IC / FUC – RS during the months of January to October 2012.

Casuistry

I1 – NBC, female, six months and 27 days, diagnosis of Down syndrome with congenital heart disease (Atrioventricular Septal Defect Rastelli type A), history of sepsis, prematurity (32 weeks gestation), hypothyroidism, renal failure, syndrome of low cardiac output and arterial hypertension, whereas the renal failure was resolved and the hypertension was controlled at the time of the evaluation. Three months and 21 days underwent to a procedure of total correction of the heart disease. It was needed an intubation for 90 days (Figure 1). As for food, the patient, since her birth was fed via a nasogastric tube without oral stimulation.

I2 – JPLC, male, six months and 14 days , diagnosis of Down syndrome with congenital heart disease (Atrioventricular Septal Defect Rastelli type A), history of sepsis, pneumonia, bronchiolitis, malnutrition and respiratory dysfunction. At six months he had the total correction of the heart disease. It was needed intubation for three days. As for the food, was offered the breast since his birth to around five months when he was called for bottle-feeding on the basis of multiple and recurrent breastfed desaturation, due to the effort to milking. Still, the patient had low weight gain, with signs of food aversion, setting a framework for malnutrition in this way, it began to receive feeding through a nasogastric tube (Figure 1).

Sample Characterization	Individual 1	Individual 2
Gender	female	male
Age (months:days)	06:27	06:14
Congenital heart disease	AVSD Type A	AVSD Type A
Associated diagnoses	sepsis preterm hypothyroidism renal failure hypertension	sepsis pneumonia bronchiolitis malnutrition respiratory dysfunction
Corrective surgery (months:days)	03:21	06:00
Intubation time (days)	90	3
Feeding history	Feedding tube since birth	breastfed up to 5 months bottle bottle + feeding tube

Caption: AVSD – atrioventricular septal defect.. Characterization of the sample protocol developed by the authors for data collection from medical records.

Figure 1 – Data relating to the casuistry, collected through protocol sample characterization with research in medical records during hospitalization period at the Institute of Cardiology of Rio Grande do Sul IC / FUC-RS

Procedures:

The operations were performed by the same speech therapist, following the conduct of the hospital.

The investigation of the swallowing process in two infants with Down syndrome and congenital heart disease involved the following activities:

1) Search records using the protocol for sample characterization to collect data regarding age, gender, cardiac diagnosis, associated diagnoses, number of corrective surgeries, duration of intubation and after extubation time to clinical evaluation and history of food.
2) Clinical evaluation of swallowing by means of an instrument for assessing the readiness of preterm infants for oral feeding – adapted (Fujinaga, 2002) [9]

Protocol for Sample Characterization

INITIALS: _____ Medical Record Number: _____

1) Gender: male ( ) female ( )

2) Age: _____ months _____ days

3) Heart disease: _____

4) Associated Diagnoses: _____
_____

5) Corrective surgery: yes ( ) no ( )

6) Corrective surgery realized at age: _____months _____ days

7) Intubation? yes ( ) no ( )

8) Intubation time: _____ days

9) Food history from birth:
_____
_____
_____

DATE: _____/_____/_____

Figure 2 – Protocol for sample chacacterization used to colect data in medical report

Instrument for Assessing the Readiness of Preterm Infants for Oral Feeding - adapted (Fujinaga, 2002)

Identification
Date:____/_____/_____ Name:_____
Medical report:_____ Birth date: __/__/____ Time: _____
Gestational age: _____ Age post-birth:_____ Corrected age: _____ Weight:_____
Tube feeding: ( ) yes ( ) no
( ) nasogastric ( ) orogastric ( ) nasoenteric volume: _____ml

Corrected age:
(2) greater than or equal to 34 weeks;
(1) between 32 to 34 weeks;
(0) less than or equal to 32 weeks.

State of Organization Behavior

State of consciousness	(2) alert	(1) light sleep	(0) deep sleep	
Global Posture	(2) flexion	(1) semiflexion	(0) extension	
Overall tone	(2) normotonia	(0) hypertension	(0) hypotonia	

Oral posture

Posture of the lips	(2) Sealed	(1) parted	(0) Open	
Tongue posture	(2) flat	(0) High	(0) retracted	(0) protruded

Oral reflexes

Rooting reflex	(2) present	(1) weak	(0) absent
Sucking reflex	(2) present	(1) weak	(0) absent
Biting reflex	(2) present	(1) exacerbated	(0) absent
Gag reflex	(2) present	(1) present anterior	(0) absent

Non-nutritive sucking (the test duration shall be one minute)

Movement of the tongue	(2) adequate	(1) altered	(0) absent
Cupping tongue	(2) adequate	(0) absent	
Adequate movement of the mandible	(2)adequate	(1) amended	(0) absent
Suction force	(2) strong	(1) weak	(0) absent
Sucking and pause	(2) 5-8	(1)> 8	(0) <5
Maintenance of rhythm	(2) rhythmic	(1) arrhythmic	(0) absent
Maintenance of the alert state	(2) yes	(1) partial	(0) not

Signs of stress: (2) absent (1) up to 3 (0) over 3

accumulation of saliva	( ) absent	( ) present
nasal flapping	( ) absent	( ) present
skin color variation	( ) absent	( ) present
apnea	( ) absent	( ) present
tone variations	( ) absent	( ) present
change of position	( ) absent	( ) present
tremors of tongue or jaw	( ) absent	( ) present
hiccup	( ) absent	( ) present
cry	( ) absent	( ) present

Score:_____ Maximum score: 36

NUTRITIONAL SUCKING
Bottle's kind: _____ Flow: _____ Beak: _____
Consistencies:
( ) Liquid
( ) Liquid-thickened ____%

Therapeutic techniques used:
( ) pauses_____
( ) sucks / pause
( ) interspersed Nutritional sucking and non-nutritional sucking
( ) others_____

Figure 3 – Instrument for Assessing the Readiness of Preterm Infants for Oral Feeding – adapted (Fujinaga, 2002)

RESULTS

The individual 1 was sent to the speech therapy service it was held a clinical evaluation by means of an instrument for assessing the readiness of preterm infants for oral feeding – adapted (Fujinaga, 2002)[9] after six days of extubation. The evaluation was performed using a gloved finger and pacifier with and without taste stimulus. As to the state of consciousness, the patient was alert, semiflexion global posture, hypotonic overall tone; regarding posture of lips were parted, laying flat tongue; rooting reflex and absent sucking, biting and gag reflexes present; movement of tongue, tongue cupping and moving jaw absent; missing suction force; remained alert during the evaluation; showed no signs of stress; presenting score 14.

Results	Individual 1	Individual 2
Days pos-extubation	6	11
State of consciousness	alert	alert
Global posture	semiflexion	flexion
Global tone	hypotonic	normotensive
Posture of the lips	parted	parted
Tongue posture	plane	plane
Rooting reflex	absent	present
Sucking reflex	absent	present
Biting reflex	present	present
Gag reflex	present	present
Movement of tongue	absent	altered
Cupping tongue	absent	absent
Movement of jaw	absent	absent
Suction force	absent	strong
Maintenance of rhythm	absent	arrhythmic
Maintenance of the alert state	yes	yes
Signs of stress	no	yes: 1
Score	14	25

Figure 4 – Results of clinical evaluation through Instrument for Assessing the Readiness of Preterm Infants for Oral Feeding – adapted (Fujinaga, 2002)

The individual 2 was subimitted to the speech therapy service, it was held a clinical evaluation by means of an instrument for assessing the readiness of preterm infants for oral feeding – adapted (Fujinaga, 2002)[9] 11 days after extubation. The evaluation was performed using the pacifier without taste stimulus. As to the state of consciousness, the patient was alert, global bending posture, overall tone normotensive; regarding posture of lips were parted, laying flat tongue; rooting, sucking, biting and gag gifts; tongue movement amended; cupping of the tongue and jaw movement absent ; strong suction; kept the arrhythmic form of sucks; remained alert during the evaluation; showed a sign of stress: drawing; presenting a score of 25 (Figure 2) . It was possible to start with oral workout with a bottle of milk with common flow nozzle, the liquid being thickened to 3% settling breaks every 3-4 sucks.

DISCUSSION

Children with genetic syndromes often have some kind of difficulty or related to feeding and swallowing, are usually resulting from the interaction of anatomical, physiological, diagnostic and behavioral factors dysfunction conditions, making the feeding process by often cornered, tiring and negative. These issues favor the development of behaviors that make eating difficult, such as refusal or oral aversion, which limits the oral motor experiences and impairs the acquisition of skills in that aspect [1,7]. The facial phenotype characteristics and oral motor development in Down syndrome show the possibility of disturbances in the swallowing process. The history of the two infants in study demonstrates the difficulty with the feeding experiment, in which the anatomical and functional conditions, so that the process of swallowing was performed safely and effectively, presented themselves changed. The deficit that this negative experience cause may have been reflected in the results of the clinical evaluation by means of low scores and nutrient intake of the individual 2, configured as “malnutrition”. Prior to clinical evaluation, the individual 2 was being fed through tube to make possible the supply of caloric intake necessary for the development of the infant and clinical stabilization, after the evaluation it was unable to start the training of oral with volume control and consistency used, and the liquid thickened to standardize thickening of 3 %. It was found arrhythmic nutritive sucking pattern requiring pauses to each group of 3 to 4 sucks.

The choice of protocol for clinical evaluation was due to be the only instrument in the Portuguese language that is validated and has adequate reliability among its observers in a similar population – infants preterm and term infants [8-10]. In its clinical validity, was recommended for newborns studied cutoff of 28 points on a scale of zero to 36 to begin the process of transition to oral feeding. Infants with Down syndrome and congenital heart disease aged six months to 14 days and six months and 27 days resembled the premature infants at low readiness scores for oral feeding and in clinical management indicated, related training of oral motor functions or volume restriction via oral[1,8-10].

The present study demonstrated that in both individuals the motion characteristics, positioning and muscle tension were altered, with lips parted as usual posture, tongue movements missing or changed, tongue cupping missing and lack of jaw movement during clinical evaluation. Previous studies performed with surface electromyography in children with Down syndrome showed they may have specific characteristics related to oral motor development, among them are: intra and extra-oral hypotonia, labial closure and control of tongue movements changed being movements of tongue and jaw fundamental for milking and coordination of sucking, swallowing and breathing and labial essential so that there is intraoral pressure essential to the effective suction [10,11].

In pediatric cardiac population, are considered risk factors for oropharyngeal dysphagia: less than three years old, greater than seven days of intubation, preoperative intubation and operations for obstructive lesions of the left side[6], corroborating the present study, in which both infants showed the symptom and at least two risk factors mentioned above.

It is worth mentioning that there are several studies that report the existence of feeding difficulties in children with congenital heart disease diagnosis and that, usually, these difficulties are characterized by denial, aversive behavior, food preference and lack of competence to their level of development[1,5,12-16] .

Similarly, orofacial miofunctional changes may also be responsible for eating disorders characterized by oral dysfunctions[15]. In this case study, the infants showed signs of swallowing difficulties characteristic of infants with congenital heart disease and signs of oral disorders that represent miofunctional changes previously described in Down syndrome. In order to corroborate the previous literature, the two infants exhibit the symptom dysphagia which may be linked to several predisposing factors: the difficulty of swallowing within syndromes that impair motor skills, to miofunctional changes in Down syndrome with congenital heart disease, corrective surgeries as well as mechanical ventilation and period of prolonged hospitalization[1,7,8,12-17] .

The present study had some difficulties related to logistics assessments of swallowing, considering that infants with Down syndrome and congenital heart disease may exhibit unstable clinical condition and intubation for a prolonged period. These conditions may prevent the achievement of clinical evaluation in a large number of infants during the period and age proposed by the study. Another limitation to be mentioned is the difficulty of performing clinical evaluations of previous corrective surgery of the heart defect, swallowing in order to verify the presence of dysphagia and predisposing the same symptoms in the pre-surgical. This occurred because infants often are intubated during hospitalization or because infants from other cities come to the hospital only to surgical intervention, since the IC-FUC/RS is a referral hospital for heart disease, apart from the fact that patients need to be referred for speech therapy service, which usually occurs

after surgery. Still, the study had difficulty in providing instrumental swallowing evaluation to describe the symptoms of swallowing difficulty objectively, since this exam is conducted by medical referral and is not routine care in a cardiology hospital.

Finally, we emphasize that although the syndrome is a widely known condition, deserves scholarly work in the area of pediatric swallowing emphasis because they are scarce. The need is evident mainly to verify the morphological, anatomical, functional and clinical status that usually affect infants these characteristics. The evaluation and early intervention can be critical to provide a better development of oral motor functions, better overall development and quality of life for infants with congenital heart disease and dysphagia.

FINAL COMMENTS

Thus, it is concluded that even infants with Down syndrome in old age may have incoordination of sucking, swallowing and breathing in the postoperative period.

The clinical evaluation showed the presence of oropharyngeal dysphagia in both cases studied. Dysphagia presented in the two cases described as a hinder action for oral feeding.

The study suggests the importance of speech therapy in heart disease and syndromic pediatric population through low scores presented, similar to preterm infants in studies that used the same protocol for the clinical evaluation of swallowing, and especially the need for the development of oral and food skills, to provide clinical quality of life and stability to them.

Given the importance of this experience for the overall development of infants, future prospective studies with a larger number of subjects are needed to contribute to the series of cases, and to verify the presence of preoperative dysphagia or demonstrate the influence of factors predisposing dysphagia.

RESUMO

O presente estudo teve por objetivo realizar avaliação fonoaudiológica da deglutição em lactentes com diagnóstico de síndrome de Down e cardiopatia congênita (DSAV tipo A de Rastelli) internados na unidade 2A e Unidade de Tratamento Intensivo Pediátrica da instituição de origem, com suspeita de dificuldade de deglutição, encaminhados ao serviço de fonoaudiologia. Trata-se de uma pesquisa de caráter descritivo-qualitativo, nessa foi possível avaliar dois lactentes durante o período de janeiro a outubro de 2012. Utilizou-se um protocolo de perfil da amostra para a coleta de dados sobre histórico clínico e diagnósticos dos prontuários dos pacientes, além do instrumento de avaliação para prontidão do prematuro para alimentação oral proposto por Fujinaga (2002) para avaliação clínica da deglutição. Os pacientes apresentaram como resultados a presença de disfagia orofaríngea e escores baixos inviabilizando a alimentação exclusivamente por via oral. A conduta terapêutica foi de estimulação oral e volume de alimentação controlado para alimentação oral. Os achados corroboram a literatura no que diz respeito à relação entre disfagia, cardiopatia congênita, síndromes genéticas e alterações miofuncionais orofaciais. Conclui-se que a disfagia apresentou-se como um sintoma dificultante para a alimentação por via oral de forma segura e eficaz para todos os lactentes estudados. Estudos prospectivos com um número maior de sujeitos são necessários para contribuir com a série de casos e, desta forma, identificar outros fatores de risco para disfagia bem como condutas terapêuticas específicas para crianças com síndrome de Down e distúrbios da deglutição associados.

DESCRITORES: Transtornos da Deglutição; Síndrome de Down; Cardiopatias Congênitas; Lactente; Deglutição; Avaliação

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Mailing address:
Deborah Salle Levy
Rua Ramiro Barcelos, 2600, térreo, secretaria – Santa Cecília
Porto Alegre – RS – Brasil
CEP: 900035-903
E-mail: deborahslevy@gmail.com