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DOWN SYNDROME: DIFFICULTIES IN TRANSMITING THE DIAGNOSIS

SÍNDROME DE DOWN: DIFICULDADES EM TRANSMITIR O DIAGNÓSTICO

SÍNDROME DE DOWN: DIFICULTADES EN TRANSMITIR EL DIAGNÓSTICO

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ABSTRACT

Objective: to identify the difficulties of health professionals in communication of the diagnosis of Down Syndrome, identifying forms of approaches used to convey the diagnosis. ***Method:*** a descriptive study with a qualitative approach, developed in a public hospital in the city of Imperatriz / Maranhão / Northeast of Brazil, with 12 health professionals. It was used a semistructured interview guide and for data analysis, it was adopted the technique of analysis of thematic content. The research project was approved by the Research Ethics Committee under number 274/11. ***Results:*** most of the professionals are not prepared to convey the diagnosis and guidance of Down Syndrome to parents, and prefer to reveal the news only in the presence of the mother. ***Conclusion:*** it is relevant to the improvement and enhancement of all professionals who deal with the diagnosis of Down syndrome, with a view to provide adequate information about the syndrome. ***Descriptors:*** Down Syndrome; Parents; Health Professionals.

RESUMO

Objetivo*:** identificar as dificuldades dos profissionais de saúde na comunicação do diagnóstico da Síndrome de Down, identificando as formas de abordagens utilizadas para transmitir o diagnóstico. ***Método*:** estudo descritivo, com abordagem qualitativa, desenvolvido em uma maternidade pública localizada na cidade de Imperatriz/MA/Nordeste do Brasil, com 12 profissionais de saúde. Utilizou-se um roteiro de entrevista semiestruturado e para a análise dos dados, adotou-se a técnica de Análise de conteúdo do tipo temática. O projeto de pesquisa foi aprovado pelo Comitê de Ética em Pesquisa sob número 274/11. ***Resultados*:** a maioria dos profissionais não está preparada para transmitir o diagnóstico e as orientações da Síndrome de Down aos pais, e que preferem revelar a notícia na presença apenas da mãe. **Conclusão:** torna-se relevante o aprimoramento e aperfeiçoamento de todos os profissionais que lidam com o diagnóstico da Síndrome de Down, com vistas a fornecerem informações adequadas sobre a Síndrome. ***Descritores: Síndrome de Down; Pais; Profissionais de Saúde.

RESUMEN

Objetivo: identificar las dificultades de los profesionales de la salud en la comunicación del diagnóstico de síndrome de Down, la identificación de las formas de los enfoques utilizados para transmitir el diagnóstico. ***Método:*** se realizó un estudio descriptivo con abordaje cualitativo, desarrollado en un hospital público en la ciudad de Imperatriz/Maranhão/Noreste de Brasil, con 12 profesionales de la salud. Se utilizó una guía de entrevista semiestructurada y el análisis de los datos, adoptamos la técnica de análisis de contenido temático. El proyecto de investigación fue aprobado por el Comité Ético de Investigación con el número 274/11. ***Resultados:*** la mayoría de los profesionales no está preparada para transmitir el diagnóstico y la orientación del síndrome de Down a los padres, y prefieren revelar la noticia sólo en presencia de la madre. ***Conclusión:*** es relevante para la mejora y el fortalecimiento de todos los profesionales que trabajan con el diagnóstico de síndrome de Down, con el fin de proporcionar información adecuada sobre el síndrome. ***Descriptores:*** Síndrome de Down; Padres; Profesionales de la Salud.

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INTRODUCTION

The journey faced by patients before a medical diagnosis can be slow and painful, especially in cases of incurable diseases, rare with poor prognosis and mutilating. In childhood, the diagnosis of disease burden in this age group parents and guardians, especially in malignancies, trauma and genetic alterations such as Down syndrome, Turner syndrome, deafness, among others.[1]

The impact of these changes in childhood will have an effect on growth, development and social relationships of children and their families, especially Down syndrome. This syndrome occurs due to an error in the division which leads to chromosomal triplication of chromosome 21 instead of duplicating this. The etiology of this change is unknown, but are known ways in which it can occur. The most common mechanism (94% of cases) occurs by non-disjunction of chromosomes, and therefore all cells end up purchasing an extra chromosome 21.[2]

Estimates indicate that every year are born more than six thousand children with Down syndrome in the United States and Latin America, with a frequency of approximately one in every 700-800 births, and in Brazil, around one in every 650-700 live births. Faced with such magnitude becomes increasingly necessary that health professionals are prepared to provide comprehensive care before this syndrome.[3]

It is known that Down syndrome is characterized by clinical signs that cause the delayed development of motor functions, such as sucking, swallowing, talking and walking. However, patients with this disease have the ability to develop daily tasks and participate in social life with the family. Despite these good estimates a child with this genetic alteration to society is still a human being with failure to maintain social relationships and starts to face prejudice and difficult situations.[4]

Given this, parents develop a concern soon after birth, following the discovery of a child with Down syndrome. Initially, they experience the shock of the news regarding the diagnosis, which can be mitigated through the way the family is diagnosed by health professionals, making it necessary to organize to support acceptance of the disease. In this perspective, the literature argues that the health care professional who meets this patient needs theoretical, practical and mental infinitely meaningful to manage the difficulties facing the family.[5-6]

The diagnosis becomes a unique moment for the family, considering that parents do not plan the birth of a child with genetic alterations, thus the time to inform the diagnosis is important for the adaptation of the family receives a child with this syndrome. The care and attention of health professionals at this time decisively influence the image that all family members will have the child.[6]

However, observing the care provided by health professionals to convey the diagnosis of Down syndrome, it is perceived to be too vague knowledge gained so far on the subject, causing many doubts and feelings that impose difficulties in conducting relevant guidelines on the syndrome with imminent risk to commit decisively in adjusting and adapting subsequent parent and child. It is noteworthy that the higher the support and emotional support given to parents at this time, the easier it is to understand the new situation that will have to live.[3,7]

Considering the need to confront this reality, the study is constituted as a subsidy that can guide care practices thus aimed

- To identify the difficulties of health professionals in the communication of the diagnosis of Down syndrome, identifying forms of approaches used to convey the diagnosis.

METHOD

Article compiled from Labour Course Conclusion << **Disclosure of the diagnosis of Down syndrome to mothers: challenges for health professionals** >> presented to the Nursing Course at the Center for Social Science, Health and Technology/ACSH, Federal University Maranhão/UFMA. Empress/MA, Brazil. In 2011.

Descriptive study with a qualitative approach, developed in a public hospital, located in the city of Imperatriz, chosen because it is the only public hospital reference southwest region of the state, which has health professionals responsible for diagnosing and monitoring children with the syndrome down. Motherhood receives daily in addition to local demand, people from other states, as the Tocantins and Pará Motherhood also has an outpatient interdisciplinary known for Follow-up for newborns with disabilities resulting syndromes, whose goal is to perform screening of children who use this service and monitor those with neurological sequelae.

The study subjects were 12 health professionals from motherhood. The eligibility criteria were: being professional healthcare acting in obstetrics, neonatology and

psychological counseling and have experience to inform and guide about Down syndrome to parents.

Data were collected during the months of November and December 2011, through interviews, and as a tool for data collection a questionnaire semistructured include variables related to the ways of transmitting the diagnosis of Down syndrome to parents, the difficulties of communicating such a diagnosis and approach used during this time. The average length of the interviews was 15 minutes, recorded in MP4 player and transcribed for later analysis.

For data analysis, we used the analytical framework thematics[8] content type, which involves a set of analysis techniques of communication in order to obtain, for systematic and objective procedures, a description of the content of the speeches, covering the pre analysis and exploration of data, followed by organizing the data into four thematic categories. To maintain the confidentiality of the information reported by the participants, these were identified by occupational category, followed by a number.

The research conformed to Resolution No. 196/96 MS[9], beginning after approval by the Ethics Committee on Human Research of the University Hospital of the Federal University of Maranhão, with opinion adopted No. 274/11.

RESULTS AND DISCUSSION

The data analyzed correspond to the categories found in this study are shown below.

- **Category 1 – Characteristics of study participants**

To obtain data from a multidisciplinary approach with the communication of the diagnosis and guidance of Down syndrome were interviewed twelve health professionals, four doctors, three nurses, two physiotherapists, two psychologists and an occupational therapist. The team has multidisciplinary character, which contributes to the family is welcome and can experience and express their feelings with appropriate professional support.

Respondents have age between 26 and 60 years old, living in the city of Imperatriz, six of which are natural from Maranhão and the other from other states in Brazil. Among the participants, only four reported having specialty in pediatrics. In addition, training time has around 3-12 years.

- **Category 2 – Training of health professionals to convey the diagnosis**

The participants defined as precarious and insufficient, the academic qualification received for the development of diagnostics that can modify the construction of living beings, such as Down syndrome, which might directly influence the quality of care provided by professionals, as can be envisioned in the speech:

> *The preparation is insufficient, because the professional need better preparation to cope with this situation, so delicate, that this diagnosis is to communicate and pass the guidelines to parents. (Nurse number 1)*
>
> *The preparation is poor. Because there are many theories on Down, but not really professional trained to address the practical difficulties of this moment, which may influence the process of initial adaptation of parents to the child. (Psychologist number 2)*
>
> *I think the preparation flawed, because you have to be prepared not only scientifically, but emotionally to give this news to relatives. (Medical number 4)*

The results of the present study were similar to others that deal with the question of the diagnosis of Down syndrome, to emphasize on the unpreparedness that most professionals have to transmit diagnostic-related genetic parents, which suggests visible flaw during preparation academic. [10,4]

Investigations on the transmission of the diagnosis of Down syndrome are gaining space in the national and international literature. Faced with this realization, it becomes essential to accomplish particular approach on how communication diagnostics, especially those that will lead to new adaptations family. Thus, it is during the academic, these issues need to be raised, to include in the curriculum disciplines that address, for example, about Down syndrome, since this requires the professional preparation not only scientific, but also building skills and extreme emotional humanization during this time.[11,7]

Confirming the findings in the literature, most professionals are not prepared, technically or emotionally, to deal with this event, and may experience some feelings of anxiety, helplessness and discomfort when having to communicate the news to the parents.[5-6,12]

Moreover, conceptions and inadequate information resulting from the lack of professional training, immediately after the child's birth, may contribute negatively to the bond between parent and child is favorable, and generates misconceptions regarding the syndrome, which can contribute so that parents do not trust the chances of your child's development.[6,13]

• Category 3 – Approach of professionals in the communication of the diagnosis of Down Syndrome

It is observed in this study that the approach used for the development of diagnostic Syndrome is unique to each health professional. Of the twelve interviewed seven prefer to communicate the diagnosis of genetic modification only in the presence of the mother. In this sense, it is possible to conclude that the option to communicate the diagnosis to the mother alone, relates to the closeness that has with the child, as the speeches that follow:

> *I prefer to talk first with her mother, in some cases, some families have already realized, but at the moment of revelation preferred to speak privately with the mother, because from my point of view it is she who deals closest to the child. (Nurse n. 2)*
>
> *Initially call the mother, she has the right to know first, explain what is the disease and the consequences throughout the child's life. (Medical n. 1)*

The literature suggests that health professionals need to be cautious at the time of diagnosis in the postpartum period, since that period, mothers experience a period of immense charm and fragility, on the arrival of the child. Thus only can discuss and understand about the meanings of the syndrome diagnosis after going through this phase.[13-4]

Thus, the mother, to learn the news that his son has Down syndrome needs support, security and trust, making it not run away from reality, but face and build something relevant to the care of the child. In this perspective, the ideal period for information on postpartum about this diagnosis is between 5 and 30 days after the child's birth, when the mother has already experienced the first contact with the newborn and initiated the formation of the bonding.[15]

Several studies suggest, therefore, in regard to families, it is essential that the mother is not alone at the time of the news, and should be accompanied by a parent or other family members. The family receiving the news after the arrival of the child should be guided and supported to care for their child and seek the most appropriate treatment.[16,6,10] The statements below justify the benefits of this approach:

> *I think the diagnosis before the child's parents is critical, one supports the other, and must be followed by work with the family, it is very important to explain nicely. (Nurse n. 2)*
>
> *I work not only with the mother, but with the family, so that along with the child are the primary subjects, so that respect is made with real constructive awareness of values and not just social imposition. (Psychologist n. 2)*

This situation confirms the importance that professionals have to inform parents about the syndrome, highlighting even as reference studies on the need for adequate language to provide parents with clear, objective and current at the time of the news because from that time, parents face the difference between the desired and son real son, being necessary to demonstrate sensitivity of the information, however, no such omission.[16-7]

• Category 4 – Difficulties in communicating the diagnosis of Down syndrome

Regarding the difficulties of professionals communicate about Down syndrome, as well as other shortcomings, it is clear, in the reports, the reduced structure emotionally in time to inform the parents, in particular, on how to deal with the reaction of parents in this moment. The testimonials below contains this understanding:

> *The difficulty is to deal with the reaction of parents. Each family reacts in a way, and about the things they talk about the disease. It is important to be prepared, just that we are not always because parents often have many questions, cry, and need straight answers and objective. (Doctor n. 1)*
>
> *With the communication of the diagnosis of Down syndrome I work with frustration, anger, rage, mother and family. I do not know how to handle this situation in different child, try to be calm, to pass knowledge to the whole family, but I never feel fully prepared to give this diagnosis. (Doctor n. 2)*

It actually discussed among the authors that health professionals experience the shock and fear of the child's parents regarding the news of Down Syndrome, as well as pain and anxiety in the preparation of such are the expectations for the future of the child, what makes it even more delicate for these professionals to address a diagnosis of such a dimension. The parents' reaction can be determined by the social meanings and beliefs about disability.[18,14-5]

According to the communication of the diagnosis, some parents also express feelings of disbelief, fear and also expressing a desire to escape the situation, pinning hopes on the possibility of a misdiagnosis of professional logo, this comes before a moment of great tension family in which parents expect him unconditional support and humanized. [19,12]

It is understandable, therefore, the importance of health professionals who deal daily with the diagnosis of Down syndrome, are able to emotionally different reactions made by the family, hardly aware that parents will quietly accept the new information, when they are still under the impact of the news, needing support, understanding and time for them to live and develop all the facts, as well as receiving the necessary information.[16]

Still, one has to consider that it is essential professionals involved in diagnostic situations that de-structure the entire household, have extensive knowledge on the disease and the process by which parents pass so that they can understand them and give them the time to assimilate the information and mainly take the necessary decisions with clarity and rationality.[8]

CONCLUSION

The results of the study allowed us to analyze the health professionals interviewed, responsible for communicating the diagnosis of Down syndrome, have no qualifications suitable for this role, demonstrating insecurity at the moment so delicate for the life of a couple when evidencing little knowledge acquired about syndrome during the academic preparation.

In this perspective, the forms of approach used by health professionals to convey the diagnosis are fragile, because of the limited knowledge we have reported, a fact confirmed when deciding to reveal preferably inform the early diagnosis of the mother, while studies on the subject are unanimous that the communication should be planned in advance to be made whole family, ensuring appropriate respect and support to those involved with the child.

It stands out as the greatest difficulty of health responsibility dealing with the reaction of the parents, since they never plan the birth of a differently child. So, knowing that the initial reaction to the news of the syndrome may be determined by the type of information received, professionals need to be aware of the way it is presented and what the attitude of those who communicate, which can influence the acceptance of the child by the parents.

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