100 - HISTORY OF LIFE OF A CHILD WITH MUCOLIPIDOSIS II: THE PARENTS FACED WITH THE NON MOTOR DEVELOPMENT OF THEIR DAUGHTER.

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Introduction

The knowledge about the meaning of the motor expression is essential to the comprehension of how important is the human movement to Man.

At birth stage, a child is subjected, in normal conditions, to a process of acquirement of certain motor patterns that are considered as characteristic motor marks of its development. These alone create an involvement in people's imaginary, namely of the parents, with incidence inclusively in the child's own life. Despite the fact that the existence of these motor marks is really very important to the parents, as they represent the acquisition of a certain level of development, so it is any movement of a new born human being. This is then considered an intrinsic characteristic of our species and accompanies the Person from its conception to its death and when this is limited for any reason, very bad repercussions will certainly occur as a result of this evidence.

In 2004, as physiotherapist in a certain health institution, we were confronted with a 7 month old child with an unknown diagnosis, but which motor patterns indicated the existence of a very serious disease. This was later confirmed to be Mucolipidosis II (MLII), a genetic disease, in which the normal motor development was seriously compromised.

Faced with this reality, it was our intention to understand how the parents of this child would react to the "non motor development" of their daughter and how they would deal with the almost absence of movement of the child. Therefore the purpose of our study was to understand the "mourning" of the parents faced with the history of life of their own daughter concerning her "non motor development".

Theoretical field of analysis

The MLII or *I-cell* disease belongs to a group of metabolic diseases designated as lysosomal storage disorder (Vellodi, 2005). It is a rare genetic disease with autosomal recessive trait, and very little available data regarding its incidence. A study from the Netherlands reported a frequency of approximately 1 in 640.000 live births (Lee and McGovern, 2003). In Portugal there is no available data regarding the incidence of this pathology.

The MLII is characterized by the intracellular accumulation of mucolipidoses in several body organs and tissues (Scheinfeld and Kark, 2005) due to a rare deficiency in an enzyme, the N-acetylglucosamil-1-phosphotransferase, product of the GNPTA gene, mapped to the chromosome band 4q21-q23 (Lee and McGovern, 2003).

The MLII is characterized by a systemic and generalized manifestation, with repercussions in the musculoskeletal system, in the neurological central system, respiratory, cardiovascular, gastrointestinal, ophthalmologic and auditive system, which severely compromises the motor and cognitive developments and for this reason there is a wide variety of characteristics shown by the patients with this pathology (Ciência Viva, 1996-2006, Lee and McGovern, 2003; Maire and Froissart, 2003; Scheinfeld and Kark, 2005).

In picture 1 we can see the characteristics that can be present in a child with MLII and the characteristics of *Maria*[1].

Weight decrease is rapid and progressive, reaching values even inferior to the ones at birth stage. Linear growth decelerates during the first year of life and ceases around two years of age. The head circumference is normally preserved (Lee and McGovern, 2003).

The motor development retardation is normally more severe than the cognitive and is characterized by a generalized hypotonia and a poor head control that prevents the child from rolling over or standing on her feet. These and the remaining characteristics impose a severe limitation to the movement. The full clinical picture is usually evident by the end of the first year of life (Lee and McGovern, 2003), which made us to start our study at the first year of age of *Maria*.

The available treatment for patients with MLII is essentially symptomatic, and beyond that offers to the child clinical support and measures aiming to improve its quality of life (Lee and McGovern, 2003). Some cases of bone marrow transplantation (TMO) (Peters, 2003) have been described with limited benefits (Maire and Frossart, 2003). The risks and benefits of the TMO must be seriously considered in the medical decision-making process (Lee and McGovern, 2003), as this procedure causes morbidity and mortality (Ellinwood et al., 2004).

Methodology

Procedures

The investigation occurred in a health institution in the area of Oporto, and its beginning was preceded by a request to the Board of Direction of this institution and to its respective Physical Medicine and Rehabilitation Service as well.

The interviews occurred during an approximate total of 75 sessions of physiotherapy that *Maria* went to between December 2004 and June 2005. The duration of the interviews was between 30 to 45 minutes, and have all been recorded with the formal agreement of the parents of *Maria.*

The realization of the interviews had to comply with a group of ethical demands recommended by Atkinson (1998), to use the "Histories of Life". The gather of information was obtained through: i) semi-structured interviews to the parents of *Maria*, ii) elaboration of a camp diary, built during the physiotherapy sessions, where all the reactions of the parents were registered which wouldn't have ever been possible with the audio recorder; iii) medical reports, iv) other data taken from the clinical history of the child.

The questions asked during the interviews were directed to confront the parents with their daughter's development and the one called as normal and thus finding their anguishes before the "non development" of the child.

The interviews were then transferred from the audio recorder to the computer. The contents were carefully listened to, and all the passages considered as being relevant and that had then been subjected to the analytical process called content analysis, were transcribed.

Treatment and analysis of the interviews

The technique used for the treatment of the information was the one called "Content Analysis" (Bardin, 1977; Ferrarotti, 1986; Vala, 1986) since it is described to be the privileged one to treat the collected material from an empiric investigation of descriptive level. It is also being frequently used in studies set upon the method of the "Histories of Life" (Poirier et al., 1995).

The "Content Analysis" has also the advantage of being able to bring to light the non structured material, as it is the case of the semi-structured interviews (Vala, 1986) used in the present study.

The function of the "Content Analysis" followed in this study was the heuristic (Bardin, 1977) which puts it on a exploratory perspective, since we didn't have any chances to confirm or deny, following a path of discovery and leave the interviews to speak for themselves.

It was effected a descriptive analysis (Vala, 1986) of the gathered material and defined the descriptive categories (Poirier et al., 1995) of the same. To build the categories, the recorded material was listened to several times, with the purpose of finding what can be designated as the crystal nucleus of information, that is, the most relevant aspects which constitute the heart of information. After detecting this nucleus of information, we fully transcribed those passages, subjecting them to the usual analytical processes.

After taking into consideration the objectives of this study, it was necessary to identify three unit types (Vala, 1986) in the transcriptions: i) register units; ii) context units; iii) enumerating units. Finally, after all this process, it was then possible to establish the categories.

Interpretative Analysis

From the analysis of the testimonies registered through the recording of the interviews to the parents of *Maria* and from the camp diary, we were able to detect 4 categories: Love/Mobility, Anguish, Mourning/non-acceptance and Hope.

Category - Mobility/Love

Sometimes from *Maria*'s parents comes the consciousness of how unbearable the situation of the baby is. Very often, the mother reports with grief that *Maria*'s development does not occur normally, that the baby has a motor delay compared with other children of the same age *"… there have been times when she raised the head a little"* (Interview (Int.) 18); *"…if Maria was normal, … she would have already been raised… she would be nearly walking"* (Int.23).

One of the dreams of *Maria's* parents is that their daughter was like "the other children". That she could hold her head, that she could sit down without any help, that she could crawl, that she could walk... The capacity of acquiring the biped position, that is, to see a son or a daughter walking is a really exceptional event in the development of a child. As Fonseca (1986:180) refers it is the "…biped station (attitude), which the anthropologists define as the most significant biological conquest of the human species". The parents of *Maria* are no exception, to them the capacity of walking is something very important: (mother): *"…may be the parents of a normal child don't give that much importance when they begin to walk, but to us is just like a dream"* (Int.59), however as the months go by, their expectations towards the motor development of *Maria* become lesser. In a first phase, the father has the expectation of seeing her walking and crawling *"I would like to see her crawling and then go after her…"* (smile) (Int.5 and camp diary (cd.)16/12), in a second phase the mother refers that *"…I know that she should have been doing that a long time ago, but…her being able to raise the head is as important to us as if she was walking!"* (Int.42).

Despite the fact that they realize how serious *Maria*'s limitations are, the parents always showed a great love for their daughter, feeling that is transmitted to us by their acts, by their words, by telling us their dreams, by making us aware of their grieves, by the perseverance of their hopes. Very often the parents show how they love the looks of their daughter, even knowing that she has the specific characteristics of a disease; however the following saying will be the most expressive one of the love the parents feel for *Maria*: (mother) *" The other day Sérgio had a dream…he dreamt that I was giving the breast to a child which face was exactly like Maria's…then we were talking… that if then God will give us a girl, she will look exactly like Maria"* (cd.26/08).

Category - Anguish

Anguish is a feeling that is present in *Maria*'s parents since they were told the disease the daughter was suffering from. Being fully aware that *Maria*'s motor development does not occur within the normal patterns, it is very noticeable the anguish felt by the mother: *"there have been times when she raised the head a bit*" (Int.18 and cd.19/01). This understanding of the limited mobility compromises the mother's hopes that she will ever be able to realize certain motor acts that are normal at her age. In another saying the father refers: *"Sometimes I want to ask a thousand things…but these are things that hurt …"* (Int.4). This anguish in relation to answers that seem to be so painful does not prevent the father from trying to understand something that is so difficult to accept. As Félix (2002:70) writes "As reasonable human beings as we are, it is only natural to be able to understand why we have to go through a certain difficulty or such an unbearable pain…"

In one of the various medical appointments with *Maria,* the mother was in contact with a 14 year child with Trisomy 10, and she became very affected with the case. She referred that *"she only weighed 11 kg… (…) and she slobbered… (…) and she couldn't talk"* (Int.19 and cd.20/01). To her the confrontation with such a shocking situation was the confrontation with a reality that appears it is probably going to occur with her daughter. The anguish expressed in her voice and her face was accompanied with tears.

Category - Mourning/Non-acceptance

Several authors describe a mourning process lived by the parents of a child with any disability and which represents the death of the "normal"/idealized and healthy child (Bromberg, 2000; Finnie, 1980; Voivodic and Storer, 2002). In the line of Drotar (1975) and Gath (1985), Casarin (1999) describes five states regarding the parents' mourning faced with a child with disability: i) the first state is characterized by the **shock reaction** with the news of the existence of a disease; ii) the second state is characterized by a **denial of the disease**; iii) the third is characterized by an **emotional intense reaction**, where feelings like sadness for the loss of the imagined baby, rage, anxiety, insecurity for the future and impotence before an unbearable situation are present; iv) a fourth state where there is **reduction of anxiety and insecurity** before the present and the future of the child and v) a fifth state characterized by the **reorganization of the family and inclusion of the disabled child.** What we have learnt is that these states did not merely and chronologically occur in the case of the parents of *Maria*. There had been an entanglement of feelings and reactions characteristic of the different states in different occasions. This is according to the line of Bromberg (2002) when he declares that the mourning process can be divided into phases, which are not rigid and definitive but are subjected to the individual differences. In the present study there was evidence of the **shock reaction**, the denial and **non-acceptation** of the disease and the **sadness** which characterizes the first, second and third states respectively. A **shock**

reaction is expressed by *Maria*'s mother (very sad): *"they said what a perfect little thing!...Then after a certain time they said she was different… they even thought something bad had happened to her"* (Int.4). The **non-acceptation** of the "idealized child's death" expressed by the parents: (father) *"they say so many things, it seems I don't believe (…) "the only thing I believe is that this is possibly affecting the organs… because she is always the same, she doesn't put on weight, …this is the only thing I believe what I see with my own eyes…"* (Int.4 and cd.14/12); (mother) *"Maria is going to be the flower girl in the wedding… so she has to start training… she has to raise the head"* (Int.52). **Sadness** is a feeling that has been very present in *Maria*'s parents throughout the months: (father) *"…sometimes I want to ask a thousand things…but these are things that hurt…"* (Int.4). Despite the fact that it is very difficult and painful to make certain questions, *Maria*'s father tries to overcome that limitation and to obtain some answers not less difficult and painful. As it is referred by Unamuno (2001:110) "Pain is the way to consciousness and it is through it that the human beings become conscious of themselves. (…) One's own consciousness is nothing but the conscience of one's own limitation".

Category - Hope

The wish of *Maria*'s parents was that they would be able to share with the daughter most of the time of her existence, as there is no cure to MLII and the length of life is limited. It doesn't matter if she walks or not …it doesn't matter if she sits or not…it doesn't matter if she holds her head or not…these are the parents' wishes, to have *Maria* always next to them. As Shakespeare says (in the Tempest, Act IV, Scene I) "We are such stuff as dreams are made on" and the parents of *Maria* continue dreaming that one day the cure for their daughter's disease will be found. And because they dream, it helps them "…to overcome the reality. They will only become useless when they take us to evasion instead of giving us strength to cope with the confrontation with reality. That is, getting out of reality"" (Félix, 2002:90). However as both Howarth and Leaman (2001:208) wrote "Having hope in something unsustainable doesn't necessarily diminishes the intensity of hope, even with the consciousness of the unsustainable". An example of this is the fact that each time their daughter manages to make any movement, their hope is revitalized: *"the fact that she raises her head is as important as if she was walking"* (Int.42 and cd.22/03). To *Maria*'s parents, her "movement is a gleam of energy, a gleam of inspiration (…)" (Fonseca, 1986:172). The feeling of hope is even more reinforced, each time anybody rebounds the favourable condition of the baby *"the pediatrician said that if she were to compare Maria to other children with Mucolipidosis, Maria "would have the cup"* (cd.01/03).

Despite the fact that all *Maria*'s motor actions represent to the parents a supply of hope, it has come to our attention that, throughout the time they became resigned with the absence of movement of the daughter: (mother), crying *"I don't know any more…I am desperate…, I don't know whether it is best for her to lay down quietly, as it is how she feels well…or bringing her here and then she cries, …becomes unhappy"* (cd.04/11).

Picture 1 - Comparison between characteristics that can be present in a child with ML II and the characteristics of *Maria*.

MLII (characteristics)	*Maria* (characteristics)
Physical - Facial Features	
High narrow forehead	×
Bushy eyebrows	×
Puffy eyelids	×
Epicanthal folds	×
Flat nasal bridge	×
Anteverted nares	×
Long philtrum	×
Thick lips	×
Gingival hyperplasia	×
Macroglossia	×
Small and long teeth	×
Musculoskeletal system	
Congenital hip dislocation	× (subluxation)
Coxa valga	×
Generalized joint stiffness	×
Claw hand deformities	
Lumbar gibbus	×
Kyphoscoliosis	×
Fractures	
Osteoporosis	
Respiratory system	
Upper respiratory infections	
Abundant nasal secretions	×
Rhinitis	
Noisy breathing	×
Pneumonia	
Bronchitis	
Otitis media	
Cardiovascular system	
Narrow coronary artery	
Rigid/narrow myocardium	
Thickening of valves and myocardium	
Cardiomegaly	
Systolic murmurs	
Ophthalmologic system	
Strabismus	×
Corneal clouding	×
Blindness	(visual acuity:7 diopters)
Glaucoma	
Palpebral ptosis	
Gastrointestinal system	
Globus abdomen	×
Hepatosplenomegaly	×
Umbilical hernia	
Inguinal hernia	
Auditive system	
Deafness	
Motor development	×
Severe delay	
Cognitive development	×
Significant delay	
Anthropometric data	
. **Weight**	
Rapid and progressive decrease	×
. **Stature**	
Nanism	×
. **Head circumference**	
Usually preserved	**Enlarged**
Radiographic Study	×
Periosteal thickness long bones	×
Tubular bones - extremities widened and short	× × ×

Picture 2 - Comparison between the normal development of a child and the development of *Maria* at the beginning (12th month) and at the end of the study (18th month**)**

		Beginning of the study		End of the study	
		Normal development¨ (12th month)	*Maria* (12th month)	Normal development¨ (18th month)	*Maria* (18th month)
Anthropometrical Data	Weight (kg)	7,500*	6,800	9,000*	6,860
	Height (cm)	75*	64,5	85 (24th month) *	65
	Head circumference (cm)	42,25*	45	63,375 (24th month) *	45
Motor Development	Dorsal Decubitus	Adopted to change the position	Preferred decubitus	Adopted to change the position	Preferred decubitus
	Ventral Decubitus	Adopted to change the position	Does not raise the head	Adopted to change the position	Does not raise the head
	Sitting down	Stabled posture	No	. Stabled posture . Sits in a small chair	No
	Standing up	. Stays standing but holding . Starts using the u.l.to the function	No	Autonomous posture	No
	Turning	Yes	With help, u. l. beneath the body	Yes	Yes
	Rolling	Yes	No	Yes	Yes (abnormal pattern)
	Crawling	Yes	No	Yes	No
	Walking	Starts toddling alone	No	Autonomous walking	No
	Manipulation of objects	. Great capacity to explore . manual preference (begins)	No	. Draws some scrawls . Holds the spoon and manages to feed herself	Plays with an object

Legend:

¨Alexander et al. (1993); Lissaver, T.; Clayden, G. (2001) Payne, V.G.; Isaacs, L. D. (1998)

* Estimated values (Mota, 1994) considering the weight, the height and head circumference of *Maria* at birth

When analyzing picture 2, we verify that the motor development of *Maria* was far behind the normal development of a child of the same age, both at the beginning and at the end of the study.

Conclusions

Our goal was to understand the "mourning" of two People, *Maria's* parents*,* faced with the "non motor development" of their daughter, through the establishment of the respective history of life giving emphasis to that parameter of the child's development.

Being fully aware of the epistemological bias through the establishment of the History of Life of *Maria* by her parents, our study became an enormous challenge. Its realization required a very profound search about a baby under our care as physiotherapist, which was realized, most of the time, in an informal environment of several sessions of physiotherapy. On the other hand our work fell upon the reactions of *Maria'*s parents, to a private and painful aspect of their child's life, the non acquirement of certain motor marks, due to the existence of a serious and fatal disease, the MLII. We wanted to understand the answers that they would give before the "loss" of the daughter with a normal motor development.

Through the used methodology we came to the conclusion that since *Maria*'s birth, the parents were expectant that their daughter would have a "normal" development, being healthy and with much love. However, after 6 months of an idealized existence of the baby, she was sent to various medical specialties, examinations and physiotherapy sessions, hovering on her the presence of a non diagnosed disease which would possibly bring threatening repercussions to her motor development.

After about a few months of great anxiety and anguish with the unknown regarding the disease of *Maria*, the parents were told that she had MLII, a rare genetic disease seriously compromising her development, namely the motor, and with a short life expectancy.

When confronted with *Maria'*s disease, feelings of non acceptance, rage and interior revolt emerged in the parents, because this was happening particularly to them. They also experienced moments of sadness and impotence before the present and an unknown future. A growing frustration occurred in *Maria'*s parents as they were being confronted with the fact that her motor development did not correspond to the actual state of the child's disease who apparently "was well" without signs of any illness. The parents lived, in this way, a conflict between the development of *Maria*'s disease and her motor development.

As they were becoming aware of the real motor limitations of *Maria*, their expectations concerning her motor development started losing ground throughout time, and this determined that they started giving gradually more importance to the motor manifestations shown by *Maria*, even if they meant to obtain of a certain achievement far behind to her age.

Because of all this, the parents started reconfiguring their expectations, each time less and less demanding, in a clear resignation towards the almost immobility of *Maria*. However, despite the fact that their hopes that *Maria* could have a "normal" development with health were vanished with time, there was something that never lost heart: the great love they feel for their daughter which reflects in the smile, in the laughter of *Maria*.

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Resumo

A compreensão do movimento do Homem e, por sua vez, do seu desenvolvimento motor, é essencial na medida que constitui uma das características mais intrinsecamente humanas.

A criança passa pela aquisição de vários marcos motores que caracterizam cada idade e que por sua vez são muito importante para os pais, na medida em que representam a evolução, mesmo ontológica, da criança.

Em 2004, na nossa qualidade de fisioterapeuta de uma determinada instituição de saúde da região do Porto, fomos confrontados como uma criança, a *Maria*, que possuía uma doença genética rara, a Mucolipidose II, na qual o normal desenvolvimento motor se encontra seriamente comprometido (Lee e McGvern, 2003).

O objectivo do trabalho é perceber o "luto" (Howarth e Leaman, 2001) dos pais perante o "não desenvolvimento motor" da sua filha. Assim procuramos estabelecer a História de vida (Poirier et al., 1995) da *Maria* através de entrevistas semi-estruturadas a seus pais, registando os seus testemunhos num gravador áudio durante as sessões de fisioterapia. Para além das entrevistas, foi elaborado um diário de campo e recolhidos dados pertinentes provenientes dos registos médicos e da história clínica da criança e da nossa interacção enquanto sua fisioterapeuta.

Os testemunhos dos pais da *Maria* foram devidamente ouvidos e sujeitos a um processo analítico denominado análise de conteúdo (Vala, 1986).

Da interpretação dos dados concluímos que os pais foram reconfigurando as suas expectativas quanto ao desenvolvimento motor da *Maria*, cada vez menos exigentes, numa espécie de resignação perante a quase imobilidade da mesma.

Palavras-chave: História de Vida; Desenvolvimento Motor; Mucolipidose Ii

Abstract

The comprehension of the movement of the Man and of his motor development is essential as it constitutes one of the most intrinsically human characteristics.

The child passes by the acquisition of several motor marks which characterise each age and that are very important to the parents, as they represent the evolution, even ontological, of the child.

At 2004, as a physiotherapist at a certain health institution in the area of Oporto, we were confronted with a child, *Maria,* who had a rare genetic disease, the Mucolipidosis II, in which the normal motor development is seriously compromised (Lee and McGvern, 2003).

The objective of the work is to perceive the "mourning" (Howarth and Leaman, 2001) of the parents facing the "non motor development" of their daughter. So we tried to establish the life history (Poirier et al., 1995) of *Maria* through interviews not directional to her parents, by registering their testimonies in an audio recorder during the physiotherapy sessions. Apart from the interviews, the information was obtained through the elaboration of a camp diary and the register of the pertinent data taken from the medical reports and our interaction whilst her physiotherapist.

The testimonies of *Maria's* parents were duly listened to and subjected to an analytical process called analysis of contents (Vala, 1986).

From the interpretation of the data we came to the conclusion that the parents have been reconfiguring their expectations concerning the motor development of *Maria*, each time less demanding, in a kind of resignation by the nearly immobility of the same.

Key-words: Life History; Motor Development; Mucolipidosis Ii

Résumé

La compréhension du mouvement de l'Homme et de son développement moteur, est essentielle parce que ça constitue une des caractéristiques plus intrinsèquement humaines.

L'enfant passe par l'acquisition de plusieurs marques motrices que caractérisent chaque âge et que sont très importantes pour les parents, parce qu'elles représentent l'évolution, même ontologique, de l'enfant.

A 2004, dans la qualité de physiothérapeute à une certaine institution sanitaire dans la région de Porto, nous avons été confrontés avec un enfant, *Maria*, qui avait une rare maladie génétique, la Mucolipidose II, dans laquelle le normal développement moteur est sérieusement compromis (Lee et McGvern, 2003).

L'objectif du travail est de percevoir le "deuil" (Howarth et Leaman, 2001) des parents confrontés avec le "non développement moteur" de leur fille. Ainsi nous avons essayer d'établir l'Histoire de vie (Poirier et al., 1995) de *Maria* a travers d'entrevues non directionnels aux parents, en registrant ses témoins dans un enregistreur audio pendant les sessions de physiothérapie. En dehors des entrevues, la recueille d'information a été obtenue a travers de l'élaboration d'un journal de campagne et de l'enregistrement des donnés pertinentes provenant des registres des médicéens et de l'histoire clinique de l'enfant et de notre interaction tandis que sa physiothérapeute.

Les témoins des parents de *Maria* ont été dûment ouïes et assujetties a un procès analytique dénommée analyse du contenue (Vala, 1986).

De l'interprétation des donnés nous avons conclue que les parents ont reconsidérer ses expectatives sur le développement moteur de *Maria*, chaque fois moins exigeants, comme s'ils étaient résignés à la presque immobilité de la même.

Mots-clés: Histoire de Vie; Dévellopement Moteur; Mucolipidose Ii