



## Case report

## The physical education and the Infantile Systemic Hyalinosis: A case report

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## ABSTRACT

The purpose of this case report is to present the case study of a child with Systemic Infantile Hyalinosis in the educational attendance specialized in Physical Education and Art (dance). The collection took place through the Teachers' Field Diary and the interview with the child's mother. The pedagogical intervention lasted 15 months and took place at the swimming and dance workshops at the Catholic University of Brasilia, with two weekly classes in each workshop lasting 30 min each. The intervention was based on Henri Wallon's theory of emotions and sought freedom of expression, body experience, and the discovery of a body marked by disease and, in many moments, disrespected in its possibilities. At the end of the intervention, mobility, and range of motion gains were observed in motor terms, which were compromised due to the instability caused by the progressive disease. However, there were significant gains regarding self-esteem, which were relevant and significantly contributed to a better quality of life of the child.

## 1. Introduction

The Infantile Systemic Hyalinosis (ISH) is a rare autosomal recessive disease caused by a mutation in ANTXR2 gene encoding a transmembrane protein involved in endothelial development that occurs on chromosome 4q21 .21 and currently there is no cure and patients are prone to die from recurrent infections (Lu et al., 2016).

The ISH has as the main characteristics the progressive joint contractures, cutaneous anomalies, severe chronic pain and generalized deposition of hyaline material in various tissues such as skin, skeletal muscle, heart muscle, gastrointestinal tract, lymph nodes, spleen, thyroid, adrenal glands as well as persistent diarrhea with enteropathy, protein loss and recurrent infections with a very short life expectancy (Casas-Alba et al., 2018; Haidar et al., 2017; Rivera et al., 2015).

Other problems associated with ISH are hyaline nodules on the medial and lateral malleoli of the ankles, metacarpophalangeal joints, spine and, elbows with progressive joint contractures, osteopenia, cutaneous abnormalities and severe chronic pain (Rajendran et al., 2015).

Despite the seriousness of the disease and, due to the rarity, there are few studies in the literature and they refer to case studies in the field of medicine (Liu et al., 2019; Mohamed et al., 2017; Haidar et al., 2017; Elenga et al., 2017).

Regarding the Physical Education field, there were no studies or researches to indicate the contribution of this kind of intervention to ISH patients who present a similar scenario. However, the Extension and Research Project in Specialized Educational Service in Physical Education and Art (dance) for people with disabilities had offered service to a child diagnosed with ISH for 15 months. The child had attended swimming and dancing activities. The project is a partnership between the Catholic University of Brasilia and the State Secretary of Education of the Federal District.

This research aims to present a case study of a child with Infantile Systemic Hyalinosis (ISH) who took part in the specialized educational service in Physical Education and Art. The study with this child may become essential so that we can reflect on the implementation of an educational intervention in Physical Education that can contribute to the

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quality of life of people with ISH and also other rare chronic disease cases.

## 2. Methodology

This study was designed under a case study which, according to Ventura (2007, p. 383), “that aims to investigate a specific case, well delimited, contextualized in time and place so that a detailed search of information can be carried out”.

To collect the information a field diary was used by the workshop teachers to daily register the proposed activities, positive and negative responses of the child. They also recorded observations and events that should be considered in the next classes. An interview with the child's mother was also carried out based on the model of life story that, according to Boni and Quadresma (2005, p. 73), it is a method where the “researcher constantly interacts with the informant. Its main function is to portray the experiences lived by people, groups, or organizations”. In this method, a topical life story was used, which focuses on a step or a particular sector of the experience in question and considering the experience of a child perceived by the mother.

The medical and psycho-pedagogical reports were used to describe the clinical scenario of the child. And the rarity of the student's clinical case and the lack of references on Physical Education studies and literature for ISH, there was no comparison with other cases.

This study was approved by the Research Ethics Committee of the Catholic University of Brasilia with protocol number 72086117.6.0000.0029 and for the accomplishment of this study, the child's mother signed the Free and Informed Consent Form, consenting to the realization of the same.

### 2.1. Getting to know the child

At the beginning of the pedagogical intervention, the child was 7 years old, presenting a picture of Systemic Infant Hyalinosis attested by the Sarah Network of Rehabilitation Hospitals. She was born by cesarean delivery at 37 weeks and 6 days pregnant, with a weight of 1,950 g, a length of 41 cm, a head circumference of 30 cm and a fifth APGAR of 9, without any evidence of any disease being perceived, remaining for 3 days in the incubator and then discharged from the hospital. According to the mother's reports, the child cried excessively when touched, especially with arms raised, but ISH was only diagnosed when she was 1 year old.

The child had several infections that led to many hospitalizations, requiring surgical interventions in the buccomaxillary region, in addition to the removal of the gallbladder. She had the stature of a baby. Her arms and legs were stunted and contracted, not extending, as if he were in a sitting position and with flexed arms. She could not move around alone without the use of a wheelchair or a walker, however, even with this instrument, she moved slowly and with difficulty. She was in constant pain, especially in the bones and skin of the anal area, but, despite all the impairments, her cognition was preserved.

### 2.2. Clinical condition

According to the Medical Report presented by the child's mother, the diagnosis is of the autosomal recessive genetic condition called systemic infant hyalinosis (OMIM 236490), having been confirmed when the child was 1 year old by the molecular DNA sequencing of the ANTXR2 gene on the chromosome 4q21.21 that revealed deleterious mutation not previously reported.

According to the report, the child has gingival hypertrophy but is able to feed without gagging and apparently without pain. The appearance of his skin was thick and infiltrated mainly in the nasogenian portion, with facial mimicry diminished by the infiltrated aspect of the facial skin. It presents an area of papular and erythematous lesions, affecting the skin of the perioral region and auricular pavilions; the neck is short and with papular lesions, with diffuse hyperemia and with a pattern of distribution

in a collar in the neck region. Flexion of the knees, hips, and elbows is observed, resulting from joint restriction and, in the hands, a restriction for a complete interphalangeal and metacarpophalangeal extension.

It also presents an evident joint restriction of shoulders and for hip extension and Abduction bilaterally, the extension of the knees and ankles with a discrete deviation of posture in valgus and equine in a symmetrical way. There is a lesion with a condylomatous aspect in the perianal region, with the consistency of soft tissues and hyperemia. Despite the systemic infantile hyalinosis, the child does not have diarrhea, which is common in ISH, but which is not mandatory in everyone affected by it.

Thye medical report says, the ICD-10 M25.6 – Joint Stiffness not classified elsewhere is also associated with the chart; IC-10 L99.8 – Other specified disorders of the skin and subcutaneous tissue in diseases classified elsewhere; and ICD-10 Q68.8 –Other congenital musculoskeletal deformities.

### 2.3. The pedagogical intervention

The pedagogical intervention lasted 15 months and happened in the workshops of swimming and dancing for people with disabilities at the Catholic University of Brasilia. There were two weekly classes with each activity lasting 30 min. During this period there were two recesses that added up to 50 days. The child also took part in two swimming festivals and two dance shows. She missed 19 of the 90 classes provided for each workshop, in these specific situations she was hospitalized or handling problems regarding her health.

### 2.4. Theoretical assumptions of interventions

The pedagogical intervention was based on Henri Wallon's theory of emotions Wallon (1971, 1981, 1975a, 1975b, 1975c) considering mainly the questions referring to the tone of the posture that, for him, "is actually constituted to provide a material basis for the affective life" (Wallon, 1975b, p. 141). According to the author, the tone consists of two functions, the first being the function of motility which corresponds to the balance of the body and the actions related to the action and body movements and the second, the tonic function, which is responsible for the disposal and manifestation of emotions and feelings.

These two functions are interconnected, and all voluntary movement will depend on a tonic contraction determining a position for its execution (motility function), protruding from a contraction to be of short duration, and very precise (tonic function) (Boato, 2013).

The tone is the basic state of tension, continuous resistance and it sticdade of tissues or organs and is the ever-present in muscle, and therefore represents a state of readiness for action. Thus, the tonus alterations may be the increase (hypertonia), decrease (hypotonia), or equilibrium (eutonia) of the action of the motility function.

The development of the tonic function in the child depends on situations of imbalance, that is when it is in the state of hypertonia or hypotonia that its tonic function will work for rebalancing, constructing schemes through which, when faced with a similar situation, it will return to its state of equilibrium (eutonia) without further effort. This adaptation of tonus contributes to the maintenance of the postural and emotional balance of the child.

According to Wallon (1971), the quality of the affective relationship established for the pedagogical intervention that it will favor the trust for the opening of communication that will allow the teacher to understand what the child expresses through her tonus and her postures that, in this becomes a clear form of communication for the child.

It is emphasized here the importance of playfulness in the proposed activities so that eutonia is maintained and the child is attentive and available to the teacher's proposals. Based on Wallon (1975c), before entering into a means of actions that subordinate it, all the activities of the child are playful, since it does them by itself. Therefore, the

proposition of activities that are not playful, can generate in the child attitudes and reactions unfavorable to learning and development.

In addition to the elements related to neuro-psychomotor development, it was sought the improvement of body image that, according to Wallon (1981, p. 37), it is "the more or less global representation, more or less specific and differentiated that she (the child) has about her own body, which is a basic element, indispensable in the construction of her own personality".

According to Bueno et al. (2007, p. 160):

For the child to develop his or her corporeity and autonomy, both the body and the world must be built. This construction needs to be done in this body and it is the structuring of the image of that body - which involves the awareness of something and the act of perceiving and being in that world - that needs to be constructed.

Such a body image has a close relationship with self-esteem, which for Rosenberg (1965) is a set of feelings and thoughts that an individual has about his or her own value, competence, appropriateness, and that leads to positive or negative attitudes towards herself. Self-esteem can be considered as an evaluation that the person makes about herself, but certainly receives influences from the gaze and opinions of others. In addition, for Mosquera and Stobaus (2006), self-esteem is not static, presenting variations depending on social, emotional, and psychosomatic events with detectable signals in several senses and degrees.

### 2.5. Pedagogical interventions

In view of the conditions of the research participant due to HIS, this proposal was not intended to seek a cure or the disappearance of the symptoms of the disease, or even the recovery of lost joint movements due to it. On the contrary, aware of the aggressiveness of HIS, what was sought was a pedagogical intervention that could provide the student with an improvement in the functionality of movements and in self-esteem, in order to lessen the unfavorable psychological effects of the disease due to the psychosocial benefits that the activity physics provides.

For the sessions, we considered the lack of specific literature for ISH cases in the Physical Education field, taking all the necessary care so the child does not feel uncomfortable due to pain caused by sudden or incompatible movements with her condition. We led a thorough observation of her motor conditions considering the motions proposed by her at the beginning of the intervention. The initial objective was to seek a state of eutonia that would allow the child a necessary balance to understand and accept the stimuli proposed by the teachers.

At the swimming classes, the child was taught individually, but, despite showing pleasure in being inside the pool, her embarrassment was evident when the teacher made eye contact, in addition to disliking when other children or teachers came near her, presenting in a defensive manner and asking for be placed with her back to the teacher. She was anxious, impatient, and hardly talked.

It is important to note that the child already had some experiences in a liquid environment, having been taken by the father to swimming pools, but in recreational situations. She was able to dive for a few seconds and move her arms and legs, although these movements were not wide enough since she did not have good mobility at the elbow, hip, knee, and ankle joints.

In this activity, the pedagogical work carried out aimed at stimulating the child's natural movements within the water, seeking to give her greater freedom to move with autonomy, which was not possible outside this environment. In addition to this stimulus, the teacher also sought to guide her about new movements aiming to acknowledge her corporal possibilities inside the water. Over time, she became more comfortable, showing herself to be more talkative, showing greater interest in the proposed activities, and even starting to suggest what he would like to do in the pool.

In the dance classes, the child was taught in a group, because it was believed in the importance of socialization and its relationship with other children, including for the development of their self-esteem.

A children's walker was used to facilitate the child's movements and participation in the proposed activities, however, sometimes, the other children would not spontaneously play with her, which led the teacher to propose work in pairs or groups. Over time, and in the face of her detachment, the other children in her class began to interact with her in a more natural way and, despite not saying much, she showed that she liked the classes through smiles, looks, and even making some comments.

We must also consider the events in which the student participated, such as two swimming festivals that are held every six months and have the participation of students from the Physical Education Course and students from Swimming Workshop, a total of approximately 120 people inside the pool, performing recreational, cooperative and interactive activities, remembering that previously, the child had not participated in similar activities, which allowed a close experience with other people.

It is also considered the fact that, in the first 9 months of intervention in the dance workshop, in addition to the regular classes, rehearsals were made for the presentation of a show that is the culmination of the work in the workshop during the year. This show usually has between 90 and 120 children and teenagers with some disability performing with choreographies that combine in a story created or adapted by the teacher of the workshop. In addition to the audience of family members, the audience is also made up of students from public schools, totaling an audience of approximately 850 spectators. Given the prospect of participating in this show, the child's commitment was considerable, rehearsing with great pleasure and detachment.

However, with the dance show approximately 15 days away, there was a need for hospitalization for surgical intervention. Faced with this situation, seeing herself being prevented from participating in the event, the child said with conviction to the mother that she would dance and, even hearing the mother repeat that she could not because of the surgery, she insisted.

According to the mother's report, when undergoing preoperative exams, she was informed of the immediate impossibility of performing the surgery due to some problems she had at the time. Upon learning of this fact, she said to her mother: "Didn't you say I was going to dance, mother?"

Three days before the show, the mother contacted the teacher who added her again into the group and she participated in the show. It is also important to emphasize that in the spectacle the character interpreted by the child entered the stage accompanied by two other personages seated in wheelchairs that were pushed by three characters who had an intellectual disability. All three were left on stage where they performed choreography and at the end, they were led to the backstage by the three characters with intellectual disability, but by the time they were to enter the stage and drive the wheelchairs, the character responsible for this child's chair did not come in and she found herself alone on the stage in front of an audience.

At that moment, surprising everyone, she began to dance and, leading her own chair with her toes on the floor, left the stage alone, a fact that commoved the whole audience.

It was realized here that, even in the face of difficulties, she participated in the spectacle, giving a special shine to it and demonstrating the capacity and conditions to dance and to present her potentialities and her intense will to live. This show was re-presented five months later and the student had the same performance on stage.

### 3. Considerations about experienced moments

Initially, it should be considered that, despite Casas-Alba et al. (2018), Haidar et al. (2017) and Soni et al. (2016), point to the low life expectancy of people affected by ISH, the child who participated in this study, even taking into account the intensity of the problems presented by him, surpassed the prognosis of death at around 24 months of life.

Regarding this child's longevity, [Youssefian et al. \(2018\)](#) consider that the variable severity of the disease in different patients has been related to the specific mutations in these patients and their location within the gene.

It is also considered the fact that there are no similar cases in the university swimming and dance workshops or in the literature, which makes it challenging to analyze the results due to the lack of parameters for comparing them. However, it is considered that, with the same methodology proposed in this study, cases of children with Autism were treated, with marked difficulties in communication and interpersonal relationships, Intellectual Disability, Cerebral Palsy and Multiple Disability with significant results in the morphofunctional, cardiopulmonary and respiratory aspects and self-esteem.

The child who participated in this study, it is also considered that in the first nine months, there was a relative psychomotor development. Regarding the development of abilities in the liquid environment, there was an improvement in the questions regarding breath control and immersion, which can be considered a great gain in face of the limitations and difficulties presented by the child, since, among other aspects, she showed greater control of anxiety and consequently a greater respiratory control and relaxation that refer to the corporal control in the water, the acceptance of the environment and the teacher within a playful environment that marks the classes in the swimming workshop.

About the dance sessions, it was possible to observe greater mobility and some improvement in the amplitude of the movements. It is also essential to consider the child's mother report regarding the pleasure shown in relation to her sessions in the two activities, especially after participating in the first dance show.

The mother said that, in addition to the fact that her daughter slept better, she also showed a desire to go to class and contentment after it, remaining motivated throughout the pedagogical intervention. These questions directly reflected her improvement in self-esteem, since she also began to communicate with the other children in the project, with her teachers and also other teachers of the university, besides having less difficulty with the looks from others.

Also according to her mother, the child was more relaxed after the sessions, besides making positive comments about the teachers, the other students, and the classes of the project. However, from the tenth month, due to the worsening of the ISH clinical scenario, the student regressed in motor aspects. Even in the dance classes, where since the beginning of the intervention she presented greater articular mobility, there was a regression. This condition of worsening of the clinical picture led even to the abandonment of the workshop's activities.

It should be noted that there is no way to disregard the seriousness of the consequences of ISH in its patients. It is a serious chronic disease with irreversible consequences that limit the life and possibilities of the person affected by it. Thus, when thinking about a pedagogical intervention in the area of Physical Education for a student with ISH, we started towards the multiple possibilities of Physical Education and not only in the directions of questions concerning the development of physiological and motor aspects.

We sought an integral man approach, focused on aspects related to self-esteem and socio-emotional development, considering the words of Henri Wallon, for whom:

There is unity between the organic and the psychic. They are not two entities to study separately and then put them in agreement. They are simultaneously expressed at all levels of development by actions and reactions of the subject and the environment, one in relation to the other ([Wallon, 1975b](#), p. 92).

It is fundamental to observe these aspects in the face of the care of children with disabilities or with rare chronic diseases, since they are more sensitive to such interventions, considering the way they are perceived by others, considering that "even if they are not traumatizing, they always disturb the exercise and development of self-assessment

capacities, thus, the functional organization of information by the nervous system" ([Vayer & Roncin, 2020](#), p. 23).

Therefore the liquid environment was chosen as a space for student stimulation in the function of the water properties that facilitate the body movements. The almost elimination of the force of gravity, due to the buoyancy in the water, helps the balance to perform movements hampered in other environments and do not force the joints that, in the case of the student who participated in the study, were compromised by ISH.

At the dance workshop, he paid attention to Wallon's words [Wallon\(1975a, p. 79\)](#) when he stated that "the movement does not intervene only in the child's psychic development and in his relations with others; it also influences your usual behavior. It is an important factor in the temper".

The fact of initially respecting her desire not to be looked at, but gradually seeking this look, was fundamental to the construction of the process of self-acceptance since it was considered the difficulty, she had in relation to the shape of her face presented due to her disease. It is necessary to consider that, due to its context, the exposure to family members is not so complex since there is an already established effective relationship, but before teachers and other children, there was an understandable vulnerability, since she saw herself with new subjects and the fear of being judged became evident. However, the way the teachers dealt with the situation allowed not only the exposure to them but also the presentation of choreography for the audience on the day of the show, that is, was able to show her whole body in action.

Thus, it is important to highlight the role of the two swimming festivals, socializing and inclusive events, where the student was with a huge group of other children and adolescents in the same environment; and two dance shows, where she had the opportunity, together with another group of children and adolescents, to perform for an audience. This points to the need to create opportunities for the socialization of people with rare chronic diseases, where they are able to live together and integrate themselves, learning and teaching together with other people and reinforcing the naturalness of difference among all.

This can be emphasized when he was alone on the stage and, without inhibition, continued to dance until he managed to drive the wheelchair behind the curtain. These facts show the development of the child's self-esteem, which showed to be empowered and able to perform in the show, which can be credited to the respectful way in which she was treated during the pedagogical intervention where she could experience, feel and move the body, without the repressions experienced in their daily lives due to the deficiency imposed by ISH.

Finally, the words of [Fonseca \(2008, p. 447\)](#) were considered throughout the pedagogical intervention, for whom:

We are not interested in safeguarding external aesthetic effects to the child, nor in adapting to 'gymnastic engagements' or 'specific sporting situations', where behavior is often confused with unintelligible reproduction, and where the movement is censored, apanage of a traditional formation of the professor of physical education.

Thus, pedagogical intervention sought freedom of expression, bodily experience and the discovery of a body marked by disease and, at times, disrespected in its possibilities, allowing the discovery of a beautiful and capable body. The quality of pedagogical relationships and the opportunities to experience her body and perceive themselves positively affected her self-esteem and consequently improved, in several aspects, her quality of life, relevant roles in the role of Physical Education teachers.

#### 4. Conclusion

In the face of chronic and serious diseases such as Infantile Systemic Hyalinosis, Physical Education in most cases has great difficulty in providing adequate care, a fact evidenced in the difficulty of finding in

the specialized literature information that can support a pedagogical intervention in Physical Education.

However, it is necessary to highlight the importance of this area in the treatment and stimulation of children affected by such diseases. Even considering that gains obtained in motor terms, such as greater mobility and range of movements, they may have been compromised due to the instability caused by the progressive disease, the gains related to self-esteem were relevant and contributed significantly to a better quality of life.

The rarity of the case makes it difficult to compare the results with the other cases of diseases and disabilities, but the good results obtained allow to open the discussion to other illnesses that are often disregarded by Physical Education due to their severity and complexity in view of the paradigms of the area in question, including allowing the replication of the study in other children with disabilities or serious illnesses, based on the use of the founding principles of the proposed pedagogical intervention, provided that individual characteristics and needs are considered.

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