



MOTOR ACTIVITY IN WATER AND FINE-LUBINSKY SYNDROME

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Abstract

Purpose. Improve motor function by exercise in the pool.

Methods. In general, there are two approaches used to study mechanical aspects of human movement: quantitative approach & qualitative approach.

Results. There was significant differences between professional swimmers and beginners movement including: breathing pattern, hand drag and stroke, fingers distances, angle of elbow and knee flexion.

Conclusions. These results have to be taken into account by coaches in order to better understand of biomechanical concepts for technical effects of training.

Key words: Water, motor activity.

Introduction

Fine-Lubinsky syndrome was described in 1993, as a very rare syndrome which entails craniofacial anomalies, bilateral hearing loss, cataract, microstomia (small mouth), central nervous system abnormalities and psychomotor delay. (P.Cole et al. 2009).

Unusual abnormalities include brachydactyly*, camptodactyly* and anomalies of the deciduous dentition. The conditions that contributed to the onset of the pathology in this specific case, are genetic recessive.

The article is about a series of water activities suggested to a 35 year old woman, aimed to improve balance, maintenance of praxis schemes, strengthen abdominal muscles, increased mobility of the lumbar tract of the spinal column, pragmatic competence and development of intentionality.

Clinical Case

The diagnostic process included a series of examinations and assessments, such as, audiometric hearing test, which results in severe bilateral perceptive hearing loss between the frequency of 55-4000 Hz. (hearing aid was fitted at the age of 6). Internist medical examination, ophthalmology check, which results in posterior polar cataract (bilateral excision surgery with the result of fair residual eyesight), otolaryngology and neurological examination, metabolic tests, high-resolution karyotype (46 normal xx), neuropsychiatric, psychological, psychomotor and speech assessments, nephrology evaluation, orthognathic evaluation (3rd class Angle, mastication deficiency).

The latest assessment of the level of adaptive behaviour, carried out by interviewing the parents,

using the Vineland Scale, shows a deviation IQ of 425, with values below average, as regards to communication and socialization. In these areas the woman had achieved a score equivalent to a child aged 1-8 years old. It occurred, however, that values were within average as regards to motor skills, and above average as regards to daily activities, areas in which she achieved a score equivalent to a 3-10 year old.

The eye-hand coordination, the fine motor skills and imitative processes, were particularly good.

(JR.Corona-Rivera et al. 2009)

On the other hand, she has great difficulties in the area of cognitive development, perception and speech proficiency. The attentive ability is also compromised and this greatly lengthens the learning process. This series of assessments brings out a clear set of symptoms attributable to the Fine-Lubinsky syndrome, which is followed by a 100% disability certification.

Activity in water

The summer* activity proposal, to be performed in the swimming pool, lasted one hour, and it was carried out every week day. Pina always showed interest and had enthusiasm to take part in the work and was very willing to follow the instructions she was given.

The exercises, so aimed to improve her motor skills, her fitness level, and help her release the stress built up, because of her hyperactivity. It also focused on improving her communication skills outside of the family environment, in prospect of the "after us" project, which that her parents hold so dear.

For this reason, the communicative interaction with the girl took on a definite connotation of affability and warmth, everytime she arrived at the Centre, to continue until the moment she entered the swimming

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pool and beyond. Usually the greeting stage included, a wave saying "hello Pina", a hug, a kiss, the request for a kiss and the message "you are beautiful".

This way, Pina felt reassured and ready to tackle the task with great serenity, feeling more inclined to the communicative exchange. It must be said however, that since the very beginning, she showed as being particularly fond of her educator, and thanks to the L.I.S. (Italian Sign Language) they immediately established a very good relationship.

It is clear that, the whole motor skill intervention was designed within the natural conjugation of the basic and essential concept of *systemic* intervention.

In other words, it is not possible to just know or acknowledge a specific disability, to study an intervention for it, to choose a method, appropriate tools and the necessary times. If, such interventions, methods, tools and times, are not interconnected with the life, space and time, that the person is living in, *without us*.

The *systemic* logic that controls the adaptive process, compels the total awareness that, there *cannot* be places for intervention, which can be separated and disconnected.

Within this system, there are no separate "rooms", where the speech therapist, or the psychomotrist, rather than the physiotherapist or the psychologist, the teacher or the rehabilitator, embrace and put forward their methodological, emotional and relational language. There is instead one only "room", where all the operators speak the same language and pursue the same goal, which is to achieve the highest degree of self-sufficiency for the individual with the disability. But there is even more. The systemic logic, responding to its intrinsic need for an overall intervention, cannot prescind from gathering information relative to the different contexts and circumstances that revolve around the individual, outside her therapeutic time, such as family, school and leisure time. The main activities identified by our intervention in water, are listed below:

improved balance, maintenance of praxis schemes, strengthening of abdominal muscles, increased mobility of the lumbar tract of the spinal column, pragmatic competence and development of Pina's intentionality**.

*During the winter time, Pina attends lots of classes, such as, Pet-therapy with "Cane Corso", Hippotherapy, Occupational Therapy, Information Technology, Physiotherapy, Speech Therapy and Augmentative Communication, also Social Integration, Music Laboratory and Activity in water.

Generally the girl has a very good relation with water, she prefers the swimming pool, rather than the sea, being a closed environment, less dispersive, so therefore more predictable. She is able to swim in deep water too, the pool where we exercised, has a maximum height of 2,5 metres. She is able to dip her head completely under the water, sometimes this

occurs accidentally, some other times voluntarily, in order to go from one lane to another, or to dive. She can also handle with confidence the presence of other people in the swimming pool, like children for example.

From the technical point of view, Pina holds a swim with her head out of the water, the legs hold the spontaneous movement typical of the breaststroke leg, the feet naturally remain in the hammer position, this is also due to a stiffness of the joints of the foot, which is caused by the syndrome. The movement of the lower limbs although, is alternate and not simultaneous and the traction movement of the arms is asymmetrical. So, while the right arm simulates the digging action backward and low, the left arm just performs a compensation motion. The asymmetry of her movement, is more evident when she has to carry teaching aids.

The workout stage

The activity was divided into three stages.

- *initial phase of access to the water and warm up*, this is the stage when Pina goes into the water and gets used to its temperature, walking along the perimeter of the pool, where the maximum height is 1,5 metres. Using floating aid items, such as a board or a tube.

The workout begins when Pina, sitting on a chair next to her educator, waits for a gesture that tells her she can enter the water (abundance of the waiting times).

Once given the signal to go into the pool, the warm up stage begins, by the gesture "look into my eyes", aimed to draw her attention, then the sign to "get the floating tube" or "the floating tablet" and the invitation, always through a sign, to walk around the swimming pool.

At the end of this first round around the pool, Pina returns the floating aid, and this action is followed by a sign of approval (applause) that makes her understand she has been good. After a few minutes (again, abundance of the waiting times) the educator will hand in another teaching aid, that will be used for another round around the pool.

- *central phase of focused workout*, this is the stage when targeted exercises are performed. They are established, based on the level of intensity, the most difficult ones are usually performed halfway through the activity, when Pina has the highest concentration.

Another important aspect to be taken into account, when planning the activity, is the tendency of Pina to store all information schematically, turning everything into a "routine". For this reason, it is important to break the patterns of exercises every so often, and set some kind of unpredictability, in order to prevent from doing the same sequence of exercises all the times.

**The reference is to the perturbing intentionality towards the surrounding environment. Such latter aspect is indeed vital, for the purpose of personal individual growth, from many points of view.

The child who perturbs, changes and modifies the environment around her, is a curious child, willing to get involved, to acquire different life experiences



without having fear of failure, to give in order to receive, and to ultimately carry out a lot of sensorimotor experiences, that will be the foundation of her cognitive development, both intellectual and more specifically physically.

The exercises performed are:

- Free-style swim (on her own or joined by her educator).

- Swimming with the aid of floating items, such as a tube, a board, one or two skittles, a ball, one or two medium sized ball in each hand, one or two tennis ball in one's hand, or in pairs with the watercircuit.

- To recuperate one or more teaching aids, thrown randomly at the bottom of the pool, using both hands and feet.

- Going underneath the watercircuit (when there are no lanes).

- Diving from the side of the pool and plunge to recuperate a teaching aid.

- Free-style swim back and forth, along the pool.

- Throws with the a ball.

- Proprioception exercises of the hands, with a big ball.

For all the exercises, the same procedure has to be followed.

Before the exercise begins:

- sign language "look into my eyes"

- sign for "take"

- sign for the teaching aid, that we are going to use, or for the action to be carried out

- sign for which direction has to be taken

At the end of the workout:

- sign for "hand me in, the teaching aid"

- sign for "you have been good"

- I caress her

- sign for "wait"

- *final phase of relaxation*, this is a stage in which Pina swims using the floating tube, following the perimeter of the pool. Once she has swum all around the pool, she follows the sign for "stop" which indicates the end of the activity. Pina approaches the swimming pool tiny ladder and gets out of the water.

In addition to performing the exercises, we also made some observations as regards to Pina's swimming speed and her heart rate range.

Speed: 30 tanks per hour. The full length of the pool is about 20/25 metres.

Heart rate range: never exceeds 130 bpm. She has fast recovery time, the range of usual workout is comprised between 110 and 120 bpm.

Apnea: 8 seconds. This is the maximum time for apnea observed, during the workout.

In regards to the teacher's presence during the activity, it starts by following Pina from the pool side and then, after about half an hour, he enters the water with her, both to swim together (exercise with the watercircuit) and also to carry out the playing exercises with the ball.

At the beginning, and for a long period of time, the educator used to stay outside along the side of the pool, while Pina was performing the activity in the water.

But during the last period, he used to go into the water too, and Pina seemed to appreciate that a lot.

She actually asked for his presence in the water, by specific requests, thus revealing a clear sign that Pina's communicative intentionality was starting to take shape.

The exercises of the intermediate stage

At the beginning, the free-style swimming and swim across the lane, were the exercises that Pina found more difficult and less entertaining, as she had to do it without any help or teaching aids. In fact, she was starting to scream, to be agitated, to bite and flap her hands in the water. Whereas now, one can see remarkable improvements, as she remains calm and seems to be a lot less irritable.

Swimming using the tube and the board, are considered as warm up and relaxation exercises, because Pina does actually relax when using such aids.

Therefore, such exercises can be very useful for those times when she lacks in concentration or is particularly nervous, in order to continue safely the activity. She uses these items to stay above the water, but still continues to drive the action, both with her legs and her right arm.

Pina considers, swimming with one or two objects in one's hand (that can be, skittles, tennis balls, medium size balls, larger balls, etc.) as a game. But technically, by performing such exercises she strengthens a lot her lower limbs. She does not even show any sign of fatigue, proving instead a fantastic ability to swim, just using her legs. When she has to carry along the large ball, she uses a technique which is similar to that of water polo, i.e. she points the ball in the desired direction, by hitting it with her hands, with small strokes. That is why, also when using other teaching aids, for example skittles or tiny balls, it is not necessary to use specific signs to let her do the exercise, one just needs to give her the items and point her which direction to take.

Swimming in pairs using the watercircuit, is useful to strengthen Pina's legs and to improve her technique. During the performance of this exercise, in fact, we realized that, when Pina has her grip on one object held steady, there is no involuntary movements "*sincinesie*" of the upper limbs, which would be caused by the propulsion of the lower limbs. So, it results a hint of simultaneous movement of the legs, which is very similar to the breast stroke swim.

At this point, it is possible to gather that, the alternating movements of the arms and legs, which are typical of Pina's swimming technique, are a consequence of the problems that she has with her left arm, used, in fact, for movements of compensation.

The exercise of recovering objects from the bottom of the pool, where the water is not deeper than 1,2 metres, was initially included, with the aim of teaching Pina how to plunge her head under the water.

But Pina did not appreciate such task, generally trying to cheat and get back the objects using her feet. Hence,



the idea to use this exercise to strengthen the abdominal muscles. So, in order to encourage Pina to carry out this recovery exercise, were used objects that she cares a lot for, such as for example her bracelets, or the ballasted circlets that are used for the swimming courses.

At the beginning she was annoyed, but afterward, being a fanatical about tidiness, she urged on picking up all the objects. Then, thanks to the exercises performed using the large ball and the use of goggles, Pina started to plunge her head also in the shallow water, and carried out this exercise in two different ways according to the requests, i.e. picking up the objects with her feet, without dipping the head into the water, or with her hands, thus diving into the water.

The sign language used to let her perform this exercise, is "to take".

In the recovery of one or more teaching aids thrown randomly into the pool, Pina obviously acts in different ways, according to the number of items, she has to pick up.

-1 object: she reaches out for the item to pick up, she grabs it with her left hand (Pina is left-handed, but when she swims she uses her left arm, as a compensation limb) and comes back to the side.

- 2 objects: she reaches the items to recover, she grabs the first one with her left hand and the second one with her right hand, then she comes back to the side of the pool, increasing the intensity of the activity of the lower limbs.

- more than two objects: Pina tends to collect all the items in a designated point in the pool, heading them all in the same direction, without grabbing them, but keeping her hands free all the times. By doing this, she demonstrates remarkable organisational skills.

The signs used to get her to perform the exercises, are the following,

- pointing at the direction of the objects

- sign "after"

- pointing at the direction to be taken, after the objects have been recovered.

One of the phases where Pina plunges her head into the water voluntarily, is when she passes under the watercircuit. This, in fact, being larger than the usual lanes, prolongs the diving time, and allows for a proper swim underwater. During this exercise the girl performs a phase of apnea lasting approximately 8 seconds. The sign language used to carry out this exercise is "below".

The dive from the side of the pool, is the second exercise in which Pina plunges her head into the water, even if sometimes this occurs unintentionally. This is done by sitting on the edge of the pool, hands joined in front of her face, she loses her balance and with a little push on her back she enters the water. During the underwater phase, Pina does not shut her eyes, and it seems like the water in her face is not bothering her, when she returns to the surface. Often, in addition to the dive, it is included the recovery of one or more

objects, or a swim up to the opposite side of the pool, in order to complete the exercise.

- sign language for "diving"

- indicating her to get out of the water

- suggesting her to sit on the side of the pool

- repeating the sign for "diving".

The exercises of throwing the ball, were introduced to make the activity more varied and less predictable, and to improve the eye-hand coordination.

They are carried out in water, about 1 metre deep. Pina does not seem to have particularly difficulties in performing such exercises, although she does not always welcome such activity as a game.

The next step, will be to be able to catch the ball in midair, which is an exercise she already performs outside of the water. Such exercise has revealed to be very important for her parents too, so now they can play with her, also in the swimming pool.

This aspect is quite significant and totally unexpected. The sign language for performing such exercise has not yet been included in Pina's vocabulary, but it consists in imitating the throw of the ball with two hands, soon after pointing the direction, or the person, to whom she will have to throw the ball.

Proprioception exercises for the hands, are performed with the large ball, with the water up to the waistline. They consist in being able to keep the ball with both hands, underwater. The deeper the ball is, the higher is the level of intensity of the exercise. Initially Pina did not understand the purpose of this exercise, that is why she did not accept it.

But then, having slightly modified it, suggesting her to let the ball pass under her legs, or just under one leg, things have improved. She manages to push the ball underwater with both hands, but with just one hand, she still cannot control the movements of the ball and give it the right boost.

Another important achievement was, wearing the goggles. Pina is a very vain girl, she loves to put her make up on, wear lots of bracelets and necklaces, but she has always been wary in regards to wearing other type of objects. So, the educator put on the goggles himself first, and then, hoping that Pina would follow the imitative process, after about ten minutes, he tried to let Pina put them on. She did not complain or showed signs of dislike. As soon as she put them on, she was told "you are beautiful with your goggles on" and was reassured.

Thanks to the exercises with the watercircuit and the circlets, Pina is learning to plunge her head into the water, in order to find objects at the bottom of the pool. Moreover, of her own free will, while she swims, she plunges her head into the water for about 6 seconds, she breathes out underwater and resurfaces to inhale.

Diving her head into the water, greatly improves the structure of her body, facilitating the performance of movements.

Outcome



The benefits of motor activity in water, for a person affected by Fine-Lubinsky syndrome, are numerous. For greater expository clearness, it was decided to separate and classify them, in two separate groups, i.e. the *immediate benefits* and the *long term benefits*.

Immediate benefits

General relaxation. Since this year, Pina has shown a remarkable improvement in regards to her attitude during downtimes, that is, when she is not busy doing any task.

Last year, soon after the activity in the swimming pool, she could relax on the couch for about 15-20 minutes, without showing any sign of impatience or frenzy. Such a long time of relaxation, was believed to be nearly impossible to achieve, during other times of the day.

- Muscular and articular relaxation. On this purpose, looking at some pictures that portray Pina's hands and feet, before and after the activity in the swimming pool. It is evident a significant relaxation, especially of the hands, whereas such relaxation is less perceptible to the feet.

As previously described, the Fine-Lubinsky syndrome, among other things, also entails camptodactyly, that is, a permanent flexion deformity of the interphalangeal articulations. So therefore, the benefits relating to muscular and articular relaxation, are even more important for those individuals, in regard to the secondary prevention of camptodactylia.

Long-term benefits

- Increased muscle tone

- Improved balance

- Improved eye-hand coordination

- Improved of the physical structure, mostly thanks to the increased cutaneous somatoaesthetics stimulation.

-Decreased episodes of stress and anxiety, both as state of mind and trait. The reduction of anxiety is particularly noticeable by the almost vanished self-harming events. Such events, usually occurred when Pina was expressing annoyance or anxiety, biting her hands or pulling her hair. Also, to the improved management of waiting times, that previously she was unable to bear.

-Rise of the threshold of fear, of physical contact. The water is often used to break down the wall, in some individuals, which is represented by the fear of the physical contact with other people or objects.

Thanks to the numerous stimulations of the skin, that simulate a continuous contact with a foreign body, different from the individual.

- Improvement of intentionality. Pina started to express her wish and intentions. Often, during the past year activity, she specifically asked her father to get into the pool, to play with her. Such aspect, represents a great change in regards to her behaviour that, till then, had always been subjected to other people's decisions.

- Improvement of the exercises performance. All the exercises carried out during our activity, are now performed with more accuracy and serenity, so the quality of work has improved considerably. This is mainly due to a greater willingness to work in close collaboration.

- Weight loss. During the summer time, between July and September, the weight has dropped by more than 2 kgs.

These, are the priority works about studies conducted in Europe in recent years:

- Molecular genetic investigation of apparently balanced translocations in patients with mental retardation and multiple congenital malformations.

Ghent University Hospital – UZGent - Belgio
Center for Medical Genetics-

-Development of novel DNA diagnostic technologies for the detection of structural chromosomal abnormalities in case of mental retardation (EFT 5467).

University of Tartu - Estonia

The Institute of Molecular and Cell Biology

- Cryptic anomalies by CGH array in children with mental retardation and ophthalmologic anomalies - CHU

Paris Seine-Saint-Denis - Hôpital Jean Verdier –
Laboratoire de cytogénétique et biologie de la

reproduction France ILE DE FRANCE – BONDY

- Identification of the molecular basis for unexplained mental retardation

Hopital Necker - Enfants Malades -

Département de génétique

France ILE DE FRANCE – BONDY

-Chromosomic errors and syndromic mental retardations

Hopital Necker - Enfants Malades - Département de génétique

France ILE DE FRANCE – BONDY

- NGFN plus : IG German Mental Retardation Network

- MR Centre Heidelberg (Project 9): Mutation screenings and functional characterisation on candidate genes

Universitätsklinikum Heidelberg

Abteilung Molekulare Humangenetik

Baden-Württemberg HEIDELBERG – Germany

- NGFN plus : IG German Mental Retardation Network

- MR Zentrum Tübingen (project 3): High resolution SNP microarrays for identification of genome imbalances and segmental UPD's. Molecular (MLPA, qPCR) and molecular cytogenetic (FISH) techniques for validation of Copy Number Variants.

Universitätsklinikum Tübingen

Medizinische Genetik Tübingen – Germany

- NGFN plus : IG German Mental Retardation Network

- MR Centre Erlangen (Project 1): Identification, validation and functional characterization of disease causing genes and proteins in patients with mental retardation of unknown cause

Friedrich-Alexander-Universität Erlangen-Nürnberg

Humangenetisches Institut – Germany



- BNE: BrainNet Europe II: European brain tissue bank
- Network for clinical neuroscience and basic research (coordination)

Ludwig-Maximilians-Universität München
Zentrum für Neuropathologie und Prionforschung
Bayern –MÜNCHEN - Germany

- NGFN plus : IG German Mental Retardation Network
- MR Centre Munich (project4): Genome-wide hybridization of genomic DNA with oligonucleotide-based arrays

Technische Universität München
Institut für Humangenetik Berlin - Germany
- NGFN plus : IG German Mental Retardation Network -MR Centre Berlin (Teilprojekt 2): Array CGH for detection of submicroscopic genome imbalances, homozygosity mapping using SNP arrays
Max-Planck-Institut für molekulare Genetik
Abteilung Human Molecular Genetics –Germany

- NGFN plus : IG German Mental Retardation Network
- MR Centre Bonn (Project 5): identification of candidate genes for mental retardation by the detection and analysis of submicroscopic CNVs
Universität Bonn Nordrhein-Westfalen Institut für Humangenetik – Bonn –Germany Nordrhein-Westfalen

- NGFN plus : IG German Mental Retardation Network
- MR Centre Essen (Project 8): genotype phenotype correlations in patients with mental retardation
Universitätsklinikum Essen
Institut für Humangenetik –Nordrhein-Westfalen-Germany

-NGFN plus : IG German Mental Retardation Network
- MR Centre Münster (Project 6): Identification of diseases causing genes in mental retardation

Universitätsklinikum Münster
Institut für Humangenetik - Germany
- NGFN plus : IG German Mental Retardation Network
- MR Centre Dresden (Project 7): Clinical characterization, molecular karyotyping and genotype phenotype correlations
Medizinische Fakultät Carl Gustav Carus der TU Dresden

Institut für Klinische Genetik
Sachsen DRESDEN - Germany
-Ricerca clinica sulle principali Sindromi associate a craniosinostosi

Azienda Sanitaria Ospedaliera O.I.R.M. - S. Anna - Struttura Semplice di Genetica
Università di Torino - Italy

- Utilità dell'array-CGH 244K nel riconoscimento di riarrangiamenti cromosomici criptici nel ritardo mentale e nell'autismo

IRCCS OASI Maria Santissima
U.O.C.di Pediatria e Genetica Medica Troina - Italy

- AGORA: Aetiologic research into Genetic and Occupational/environmental Risk factors for Anomalies in children

UMC St. Radboud - Universitair Medisch Centrum St. Radboud – NIJMEGEN

Afdeling Kinderurologie – Gelderland -PAESI BASSI
- AGORA: Aetiologic research into Genetic and Occupational/environmental Risk factors for Anomalies in children

UMC St. Radboud - Universitair Medisch Centrum St. Radboud

Afdeling Epidemiologie, Biostatistiek en HTA
Gelderland NIJMEGEN- PAESI BASSI

- High definition genomic screening of candidate genes for neurodevelopmental disorders

Hospital Universitario La Fe (Campanar)
Unidad de Genética y Diagnóstico Prenatal

Valencia – Spagna

- NGFN plus : IG German Mental Retardation Network
- MR Centre Erlangen (Project 1): Identification, validation and functional characterization of disease causing genes and proteins in patients with mental retardation of unknown cause

Universität Zürich - Institut für Medizinische Genetik - Suisse

- Unbalanced chromosomal anomalies detection in patients with mental retardation and/or dysmorphism by CGH-Array (oligonucleotides chip - Agilent 244K): Geneva - Lausanne collaborative study

Centre Médical Universitaire - CMU
Laboratoire de Cytogénétique Conventionele-Geneve Suisse Romande

- Unbalanced chromosomal anomalies detection in patients with mental retardation and/or dysmorphism by CGH-Array (oligonucleotides chip - NimbleGen): Geneva - Lausanne collaborative study

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