

Physiotherapy for children and adults living with juvenile CLN3 (Batten) disease

Please note: the information contained in this document is intended to provide supportive guidance to families, carers and associated professionals. It is not intended to be, nor is it, medical advice for individual children or adults with juvenile CLN3 (Batten) disease. Parents and carers should consult the person's GP prior to changing medication, medical treatment or prescribed activities. If you are a professional and you require more information, in the first instance please contact Sarah Kenrick (contact details at end) who can provide you with a link to the physiotherapist for Heather House.

Every person living with juvenile CLN3 (Batten) disease is different and has different degrees of symptoms, although all of them have in common the gradual loss of the ability to see, move, eat and speak. In our specialist activity centre at Heather House we support all people with juvenile CLN3 (Batten) disease in a person centred way, so everyone has their own physiotherapy programme. All people living at Heather House have a minimum of two physiotherapy sessions per week (one in the gym and the other, if the person wishes, is in the hydrotherapy pool). The physiotherapy sessions are enhanced by the support, knowledge and experience of the physiotherapy assistants and carers working in the home and centre.

We see physiotherapy as an instrument to motivate people to be active and independent. It has to be fun, stimulating, empowering, confidence building, relaxing, relevant and always adapted to the person's needs.

Our values incorporate the following:

- The treatment is not only directed at a muscle, but also takes into account how the movement is organised in the brain by activating cognitive processes: perception, attention, memory, comparison, speech – language, reasoning, imagination, etc.
- Comparison is an important mental process to acquire knowledge, improve the consciousness, ease learning and to trigger the organisation of the system
- To improve the quality of an action, it's necessary to activate the cognitive processes that are in charge of this brain organisation
- The conscious experience can modify the biological structure; better biological structure (like better proprioception) can ease an action
- We look at the body as a receiving surface, which is why perception is so important
- Recovery / maintaining physical skills is a way of learning
- We "know" because we "move" – movement gives knowledge

To summarise the physiotherapy treatment that we provide, we could differentiate three groups:

A) Children and younger adults who are physically independent with good motor and language skills.

Physiotherapy goals: BE AS ACTIVE AS POSSIBLE.

Physiotherapy treatment: Based in the use of physiotherapy and standard or adapted gym equipment, lots of balance and cardiorespiratory exercises, use of treadmill, weights machines,

rowing and cycling. Sometimes also Rebound Therapy and horse riding (hippotherapy). Goals with prizes or merit awards aid motivation to achieve.

B) Older children and younger adults who are able to stand and walk using the support of equipment. Speech, eating and drinking, fine and gross motor skills start being affected.

Physiotherapy goals: MAINTAIN and IMPROVE the activities they are able to do. BALANCE while standing and walking. BODY AND SPATIAL AWARENESS.

Physiotherapy treatment: Mainly inspired on Therapeutic Cognitive Exercises (inspired by the Perfetti Method) and other methods like Bobath, pilates and traditional physiotherapy rehabilitation exercises.

We use parallel bars for walking and we make lots of exercises in the standing position to improve balance skills. We also have exercises sitting on the wedge of the plinth, without back support and we promote the use of the steps.

We continue to support the use of adapted gym equipment to maintain strength and respiratory and cardiac effort, but always within the person's own scope on the day. At all times we monitor and use judgement in the level of potential for fatigue; we encourage only a few repetitions, say 5, then a break. More repetitions are validated with hearty praise, always recognising the motivation that comes from others – at this stage the person may be completely blind, so our affirmations help the person feel the sense of achievement better. We always end with energy in reserve, which enables the person to function during the rest of the day and see physiotherapy as something that is achievable and enjoyable.

In this stage we start introducing the use of the seat-to-stand aid, as this gives the ability to stand for longer periods, maintaining leg and core strength and familiarise the use of the aid in a positive way, when it is needed in the future.

An example of a cognitive exercise to improve balance while standing and walking could be:

Standing up barefoot, with hand support if needed (hand on parallel bars for example). Using a 'mat' of different textures, try to find differences in the materials / carpets presented under the person's feet. We encourage not only to find differences, but also to use memory and to compare with the previous materials, by asking questions like "which one is softer? Which one feels like plastic? Can you tell what it feels like?". This helps a lot to improve attention on a task and to concentrate on the body. It also increases the perception skills of the foot receptors, so this kind of exercise can have a positive knock-on effect on the individual's gait.

C) Younger adults and adults who are no longer able to bear their own weight unaided and are fully dependant on a hoist for transfers and on staff for positioning. Most people in this group require high levels of skilled support with eating and drinking and are likely to have a PEG to provide supportive nutrition. Language skills are based more on yes/no questions and the ability to say some single syllable words.

Physiotherapy goals: COMFORT in different positions (on their tummy, standing, sitting in a different place). PREVENT pain due to a lack of movement.

Physiotherapy treatment: Focused on encouraging individuals to be active in the activities that they have learned and enjoyed most, for instance if someone loved cycling, we would offer this using

passive foot pedalling using a mini peddle exerciser. We also provide repetitive passive movement in the pool (if hydrotherapy is tolerated), massage, chest physiotherapy and other exercises that enable more effective respiratory effort.

At this stage the person will be losing or have lost the ability to initiate, or they find it very difficult to actively participate, so lots of support from people or staff who are not only familiar to them but who make them feel safe and secure is beneficial. We find that the perception of what is safe to the person changes as they age and we have to work within the person's own boundaries or parameters all the time. We provide spine global mobilisations to relax the person's back and continuous repetitive movement when working in the pool, to help the cardiorespiratory and digestive systems work better.

We try to prevent pain, discomfort and chest infections and provide high quality person centred care and support, spending time doing the things the person has always enjoyed. We encourage the person to feel that "they can", although they need a lot of support.

Example of the physiotherapy programme of someone in this group of people: Julio doesn't have the ability to move or change position by himself. He uses the hoist for all transfers. He is wheelchair dependant. He can not maintain balance in a sitting position without support. We know he likes being fit and strong. He loves challenges and feeling that he can and he has done it. The treatment is based on encouraging him to:

- Sit on the edge of the plinth with the support of two/three therapists. Once sitting, Julio needs a lot of reassurance as his spatial awareness is affected. Through communication, telling him how his body is positioned in space (you are sitting on the plinth, your feet are on the floor, your legs are bent, your arms are by your side and your head is in the middle). Helps him to feel secure and the anxiety and fear disappear.
- Stand with a seat-to-stand aid. We specifically use the Evolv Standing Frame. Julio loves standing.
- Change position from lying on his back to his side (this has a helpful impact when supporting Julio for personal needs).
- When lying on his back or sitting on his wheelchair, we encourage Julio to work with his arms, giving him weights to lift or balls to throw as he has always been proud of being strong.
- The pool is the best environment to provide continuous repetitive movement to activate cardiorespiratory and digestive functions. Also to provide joint movement and therefore stimulate the production of synovial fluid (joint lubricant) to prevent stiffness and pain. Therapy in the water also helps a lot to provide movement to the pelvis and the spine.

It is very important to support people into a standing position (using standing aids), as long as they show that this is enjoyable, to prevent osteoporosis and to aid sensory perception and spatial awareness. It also increases the pulse rate, breathing and digestion/gut activity. There are many benefits, including aiding in the prevention of chest infections, helping to maintain circulation, providing a change of position and promoting someone's sense of self.

In the day activity centre at Heather House all the people living with juvenile CLN3 (Batten) disease join together once a fortnight in a "relaxation session", which has been designed to provide better body and spatial awareness, to control anxiety and fear of movement and to find a moment to relax and focus on themselves and their friends. These sessions have shown to be also very helpful for the staff supporting people with juvenile CLN3 (Batten) disease with personal care, as movement and changing the body position is essential.

Poor spatial awareness is something to consider as a common symptom and we can find this **lack of spatial awareness** in all groups A, B and C listed above. It is very difficult to detect the lack of spatial awareness when they are more physically and verbally able, but we have found in this group of people that sometimes they don't know where 'forwards and backwards' are, or where the middle is or if their legs are bent or straight. We also think that a lot of the uncontrolled movements that they have are due to a loss of spatial awareness, because as soon as they have something (like a flexible strap to hold or a weighted blanket) to provide feedback it seems that it provides that understanding of where their body is in relation to everything else, and then they stop having that level of uncontrolled movements.

As said at the beginning, everyone is different and so are the symptoms of the disease for each person. Some people will have uncontrolled movements of their limbs, others may have tremors or small spasms; others have co-activation of the muscles when doing repetitive movements. From a physiotherapy point of view it's difficult to use relaxation or exercise alone to influence these spasms and tics. Medications to aid muscle relaxation and treat epilepsy are also required along with regular (2 hourly) repositioning when the person cannot move themselves.

Many adults tend to lose the ability to hold their head in an upright position (more so in the later stage of the disease) so their chin is almost all the time next to their chest. These individuals find it very hard to lift their heads up, and at this stage they will also have complex dysphagia so corrective therapies need to be conservative to prevent aspiration pneumonias. We haven't found a complete solution for this yet, but we try to prevent this situation by educating them in which is the best position for their heads. It could be that this is a spatial awareness problem combined with the loss of abilities, fear and weakness of the muscles.

We have also observed that when the ability to walk is decreasing, people with juvenile CLN3 (Batten) disease tend to have a Parkinson gate: small steps and flexed position of the body; and in general all of them have bradykinesia (not only when moving but also related to cognitive skills). This is why 'time' is key for the person. They need time to understand; time to process the information; time to move their bodies, etc. That's why communication is so important. Success in their physiotherapy treatment depends on the communication skills of the staff supporting the person. Everything has to be fully explained, making sure the person is able to listen, focus and understand what we are going to do and what is going to happen.

As with any neurodegenerative disease, there is a susceptibility to spasticity, muscle and tendon shortening and loss of functional range of movement in the later stages of the disease. This could therefore be seen as the norm, that the person becomes more reliant on their bed and that specialist seating is less of a priority. However, we have to understand each individual, who they see themselves as, how they adjust to their losses, and how, as for most young people, they have a desire to contribute to and experience their lives in a great way for them.

In general we do not see a loss in the range of movement the person is capable of, although we move from initiated to passive exercise. Nor do we see spasticity or rigid muscles when we work with people from ability through to loss of ability. Our view is that, whilst this can be seen as part of the juvenile CLN3 (Batten) disease process, it shouldn't be, as prevention aids physical and psychological comfort, promoting motivation and dignity.

For more information, advice or support please contact

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