**Basis of Research Project: Duchenne Muscular Dystrophy, the North Star Ambulatory Assessment, and Research Initiative**

**Duchenne Muscular Dystrophy**

The data that we shall be working with as part of the project is from subjects who have varying severities of Duchenne muscular dystrophy (DMD). DMD is a genetic disorder that is characterized by progressive muscle generation and weakness and is caused by the absence of dystrophic, which is a protein that helps keeps muscle cells intact [17]. This leads to increasing levels of disability and is a progressive condition, meaning that it gets worse over time. It’s classified as a rare disease, with around 2,500 patients in the UK and an estimated 300,000 sufferers worldwide [18]. There are currently no known cures for any form of muscular dystrophy (MD), though there are treatments available to help manage the conditions [16].

DMD is one of the more severe forms of MD and generally affects boys in their early childhoods, and those with the condition generally only live into their 20s or 30s. The muscle weakness starts in early childhood and symptoms are usually first noticed between the ages of 2 and 5. The weakness mainly affects the muscles near the hips and shoulders, so among the first signs of the disorder are when the child has difficult getting up from the floor, walking, or running. The weakness progresses to eventually affect all muscles used for moving and also those involved in breathing and the heart muscle. Many are confined to a wheelchair by 12 years of age, with those in their late teens generally losing the ability to move their arms and progressive problems with breathing.

With the aim to increase the life span and movement options of sufferers of DMD, a range of treatments are available. These range from steroids to increase muscle strength, physiotherapy to assist with mobility, and surgery to correct postural deformities. And thanks to advances in cardiac and respiratory care, life expectancy for sufferers is increasing and many are able to survive into their 30s and 40s with careers and families, and there are even cases of men with the condition living into their 50s.Additionally, there are is ongoing research looking into ways to repair the genetic mutations and damaged muscles associated with various forms of MD.

**The North Star Ambulatory Assessment**

The North Star Ambulatory Assessment (NSAA) is a 17-item rating scale that is used to measure functional motor abilities in subjects with DMD and is generally used to monitor the progression of the disease and treatment affects. The tests are to be completed without the use of any thoracic braces or any equipment assistance that may help the subject complete the activities. To carry out the assessments, the assessor conducting the assessments needs a mat, a stopwatch, a box step, a size-appropriate chair, and at least 10-metres pathway [19].

To carry out the assessment, the assessor gets the subject to carry out 17 sequential tasks. Each task is graded as follows: ‘2’ if there is no obvious modification of activity, ‘1’ if the subject uses a modified method but achieves the goal independent of physical assistance from another, and ‘0’ if the subject is unable to complete the activity independently. The 17 activities involved in the assessment with the requests to the subject are given below:

1. **Stand**: “Can you stand up tall for me for as long as you can and as still as you can?”
2. **Walk**: “can you walk from A to B (state to and where from) for me?”
3. **Stand up from chair**: “Stand up from the chair, keeping your arms folded if you can”
4. **Stand on one leg – right**: “Can you stand on your right leg for as long as you can?”
5. **Stand on one leg – left**: “Can you stand on your left leg for as long as you can?”
6. **Climb box step – right**: “Can you step onto the top of the box using your right leg first?”
7. **Climb box step – left**: “Can you step onto the top of the box using your left leg first?”
8. **Descend box step – right**: “Can you step down from the box using your right leg first?”
9. **Descent box step – left**: “Can you step down from the box using your left leg first?”
10. **Gets to sitting**: “Can you get from lying to sitting?”
11. **Rise from floor**: “Get up from the floor using as little support as possible and as fast as you can (from supine)
12. **Lifts head**: “Lift your head to look at your toes keeping your arms folded”
13. **Stand on heels**: “Can you stand on your heels?”
14. **Jump**: “How high can you jump?”
15. **Hop right leg**: “Can you hop on your right leg?”
16. **Hop left leg**: “Can you hop on your left leg?”
17. **Run (10m)**: “Run as fast as you can to….(give point).”

The NSAA assessment has been shown to be a quick, reliable, and clinically relevant method to measure the functional motor ability of ambulant children with DMD, and is also considered to be suitable to be used in research. It has also been shown to have high intra-observer reliability and high inter-observer reliability provided adequate training is provided. This means that NSAA is generally fairly reliable to be used as part of research assuming adequate training is provided to assessors. Furthermore, the hierarchy of items within NSAA were shown to be supported by clinical expert opinion, with items in the NSAA assessment being listed based on their level of difficult which is agreed by most experts, while a questionnaire-based study showing that clinicians considering NSAA as clinically relevant [21].

**The KineDMD Research Initiative**

The project undertaken as described in this report is part of a wider research initiative known as the ‘KineDMD’ study, conducted by Imperial College in collaboration with Great Ormond Street Hospital (GOSH). The study involves in excess of 16 DMD patients and 10 healthy control subjects to participate for 12 months, who are assessed wearing a sensor suit on selected days during clinical assessments at GOSH, along with using fitness tracker bracelets in the form of Apple watches throughout the trial, which collect data of everyday movements while the subjects are at home or school. A broad aim of the research initiative is to make use of AI to make sense of the data patterns collected from the suit and watches for each of the subjects and, from there, would aid doctors in being able to monitor disease progression with more precision [22]. The initiative has been futured with £320,000 through the Duchenne Research Fund to develop and test the bodysuit that captures the motions of subjects suffering with DMD [23]. The hope is that insights found would cut down the time taken to test new treatments and thus drive down the costs of future clinical trials.

The hope is that insights found would cut down the time taken to test new treatments and thus drive down the costs of future clinical trials. A further aim of the initiative is that the developed suit and associated AI techniques and research projects undertaken will help determine whether any new treatment regimes are working, which would be able to help inform doctors on future treatments. This is particularly useful for specialists, as the condition can be difficult to treat as it progresses slowly and each subject responds uniquely; furthermore, many of the assessments are done by ‘eye’ instead of using measurement and objective methods. The initiative hopes that bring AI techniques into the assessments will take a lot of the human fallibility elements out of the assessments and give a more informed perspective that is better able to understand the progression of the condition in the subjects.