

# Sport activity in neuromuscular diseases: From a case of a CMT type 4A Paralympic swimmer to a proposal of survey study on sport benefit perception

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## INTRODUCTION

Sport activity is a valuable tool to improve sense of wellness, quality of life and to break down social barriers of discrimination for disabled individuals. Apart from the issue of autonomy of the individual, many physical, psychological and social benefits arise from sporting participation, which can translate into reduced health-care costs [1].

We report here a wheelchair-bound woman with CMT type 4A who became a Paralympic champion swimmer. Longitudinal observation allowed to demonstrate, after five years of intensive aerobic physical exercise and sprint distance swimming competitions with many wins, improved proximal muscles strength with increased ability to propel wheelchair independently, enhanced quality of life (QoL) and self-esteem, remarkable improvement of depression, reduced trait anxiety.

## CASE REPORT

She started to walk at 18 months. At three years of age, she had frequent falls, claw hands and a steppage gait. By 14 years of age she developed bilateral vocal cord paresis. Her walking progressively deteriorated and at age 25 she started to use one stick and at age 28 became wheelchair-bound.

She was homozygous for a c.173\_174 insA mutation in the GDAP1 gene, determining the introduction of a premature stop codon (p.P59AfsX3).

At age 32, she was not able to swim, but was persuaded by a close friend to attend a swimming pool. She started to develop a real passion for swimming and progressively increased her workout from 25-50 m to 1200-1500 m in each pool session of approximately 1.5 hour duration, four times a week. In addition, physical training included two-three sessions of weight and aerobic exercise per week, of 90 minutes duration each, in a gymnasium. She started to play in national Paralympic competitions. As a swimmer with severe physical disability, she was classified in S3 category (range 1-10, with 1 corresponding to the most severe type of disability).

**She gained the following positions at Italian Paralympic Games:**

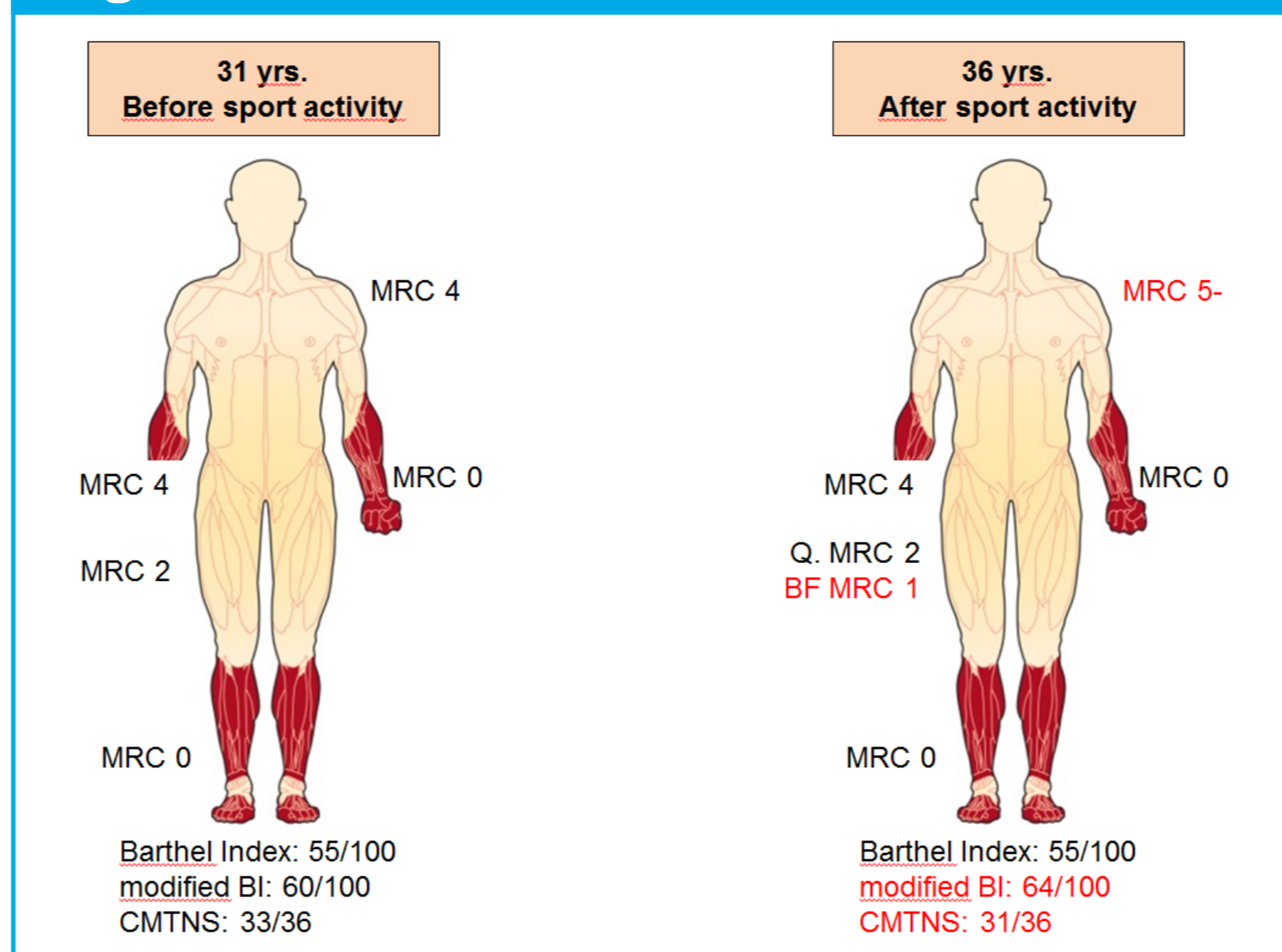
- **In 2013:** silver medal, 50 m backstroke in winter; gold medal, 50 m backstroke (Italian record) and silver medal, 50 m backstroke (category record) in summer.
- **In 2014:** gold medal, 50 m backstroke and gold medal, 50 m freestyle in winter; gold medal, 100 m backstroke in summer; 4th position in 50 m backstroke at International Championships, Berlin, Germany.
- **In 2015:** gold medal, 50 m freestyle; bronze medal, 50 m backstroke, in winter (Fig. 1).

Figure 1



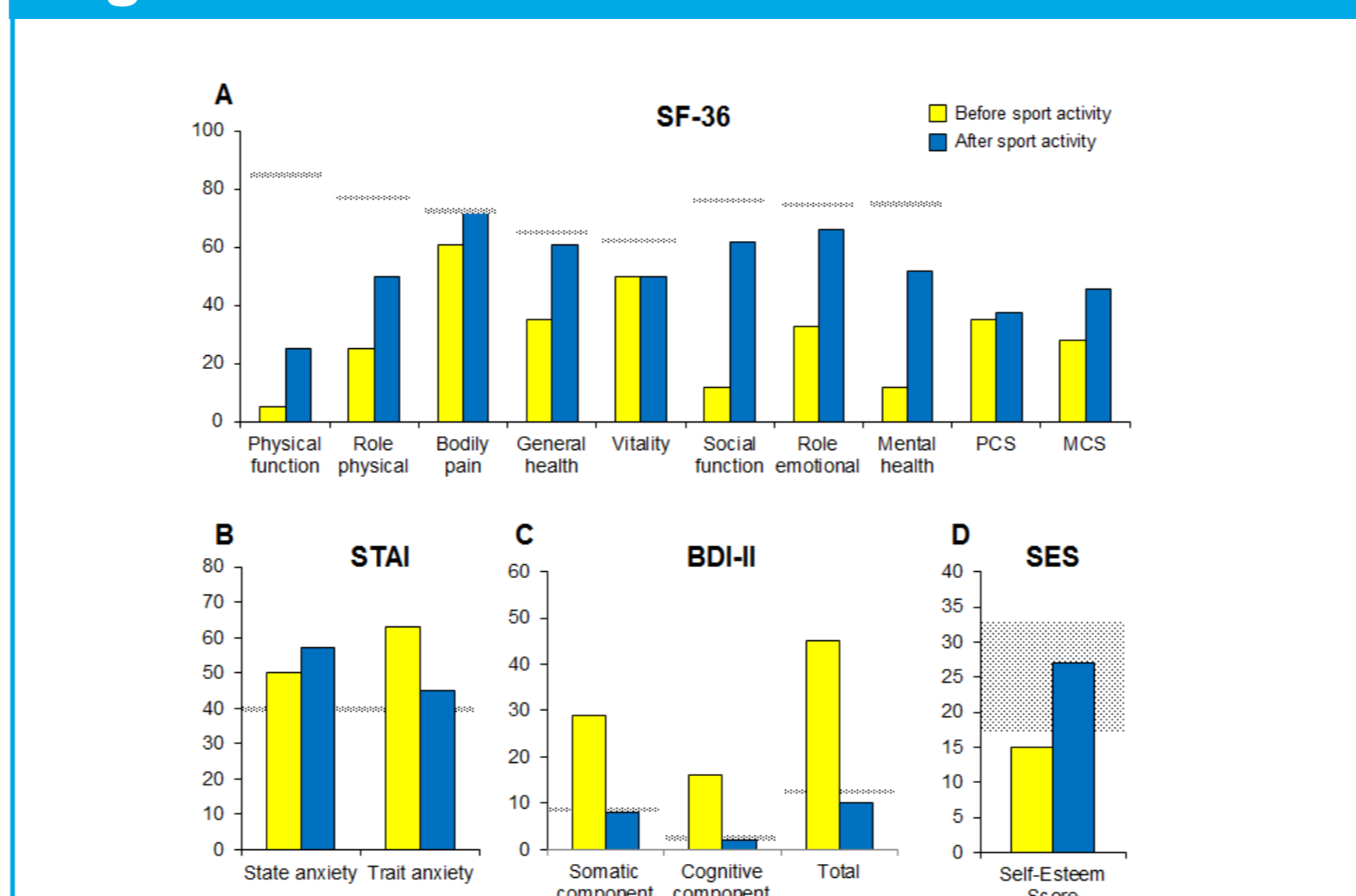
In 2015, at age 36, after 5 years of intensive sport activity, neurological and psychological examinations were compared to those at age 31 before initiating sport. We noticed an increased muscle strength in deltoid, biceps and triceps bilaterally (MRC grade 5-), whereas biceps femoris muscle strength decreased to grade 1. CMTNS diminished to 31/36 with reduced score from 4 to 3 in the items "Motor symptoms (arms)" and Strength (arms)". Barthel index (BI) was unchanged (55/100) but mBI increased to 64/100 with improvement in the wheelchair ambulation (from almost total dependence from others except short distance on flat surface to ability to propel wheelchair independently at least 50 metres) (Fig. 2).

Figure 2



Psychological evaluation revealed an improvement of all SF-36 domains except for a stable vitality. State anxiety, as transitory emotional state, slightly increased but trait anxiety, including feelings of apprehension, tension and worry as stable personality trait during daily living activities, decreased almost reaching the normal cut-off. BDI-II showed quite decreased levels of pessimism, past failures, punishment and guilty feelings, self-dislike and worthlessness with presence of ups and downs. Self-esteem returned to normal range (Fig. 3).

Figure 3



(A) SF-36 domain results before initiating sport activity and after five years of swimming activity. Pointed line indicates the mean of the Italian normative sample. (B) Stait-Trait Anxiety Inventory (STAI) score before and after sport activity. A higher score indicates greater anxiety, with a cut-off of 40 (pointed line). (C) Beck Depression Inventory II (BDI-II) score before and after sport activity. The pointed line indicates normal cut-off per each score. (D) Rosenberg Self-Esteem Scale (SES) score before and after sport activity. The pointed area delineates the normal self-esteem range.

Finally, revised form of Behavioural Regulation in Exercise Questionnaire (BREQ-2) demonstrated a strong self-determination as a result of high intrinsic and identified motivation regulating exercise behaviour. Although her physical problems, she exhibited strong self-determined motivation, suitable to engage her in high levels of physical activity. Patient perceived increased self-esteem and self-efficacy as a consequence of sport events, which were experienced as supporting her autonomy and promoting her competence.

## DISCUSSION

A recent systematic review to evaluate benefits and risks of exercise in CMT showed that the optimal exercise modality and intensity, and the long-term safety remain still unclear. However, it appears that exercise in CMT may be effective in improving some components of health and fitness without harmful effects in the short-term [2]. Moreover, we found in CMT1A no effect over time resulting in greater weakness in dominant muscles with increasing age or in more severely affected patients [3].

Most neurologists encourage physical activity in CMT patients, but recommend aerobic exercises at a submaximal work level. However, which is the best training to adopt (e.g. endurance? explosive strength?) in CMT patients is not known. We have recently reported that several StepWatch™ Activity Monitor outputs, all reflecting the higher speed, were significantly related to the main score of physical aspect of QoL: the higher the explosive performance, the better the physical QoL [4].

Although it is an anecdotal observation, the present case study leads to two main comments. Our patient, severely affected by CMT, not only was able to carry out regularly intensive aerobic swimming workout with progressive increase of covered distance, but also she competed in sprint distance events, in which anaerobic activity is prevalent to endurance, with many wins in national championships. After five years of sport activity, proximal muscle strength increased at arms with improved mBI and CMTNS, and increased ability to propel her wheelchair independently.

The second conclusion of our report is that such an intensive muscular training induced a marked improvement of QoL, removal of depression, and reduced trait anxiety. Thanks to sport practice, the patient experienced increased self-esteem and self-efficacy leading to interpret existing situations as more autonomy promoting and to organize her actions on the basis of personal goals and interests rather than controls and constraints.

Based on this case report, we propose to perform the SPORT-NMD survey study in order to investigate, through ad hoc self-administered questionnaires, the patient and family/caregiver perspective on sport and perceived benefit from it. To develop the study design, the Group will develop a protocol on the most relevant questions about patient's/caregiver's perspective on sport. NMD patients will be enrolled using different sources: clinical and genetic records of in and out-patients of third level hospitals and patients associations. Results from these questionnaires will be analyzed in order to acquire information on sport as a complementary therapy and to support provision of evidence-based advice to patients and families. Furthermore, longitudinal studies on effect of chronic training on metabolic demand, heart rate variability and biomarkers will be planned.

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