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Improving functional skills and physical fitness in children with Rett syndrome

M. Lotan,[1] E. Isakov[2] & J. Merrick[3]

1 *Zvi Quittman Residential Center, The Millie Shime Campus, Elwyn Jerusalem, Israel*

2 *Orthopedic Department, Loewenstein Rehabilitation Hospital, Raanana, Sackler School of Medicine, Tel Aviv University, Tel Aviv, Israel*

3 *National Institute of Child Health and Human Development, Office of the Medical Director, Division for Mental Retardation, Ministry of Social Affairs, Jerusalem, Zusman Child Development Center, Division of Pediatrics and Community Health, Faculty of Health Sciences, Ben Gurion University, Beer-Sheva, Israel*

Abstract

Background To investigate the feasibility of a physical exercise programme with treadmill for persons with Rett syndrome (RS) in order to promote fitness and health.

Methods A daily training programme on a treadmill was designed for four females with RS over a period of 2 months with tests performed in three intervals, at time 1, 2 and 3, 2 months apart with intervention taking place between tests 2 and 3. Participants were four girls with RS aged 8.5–11 years (mean: 10 years) attending the educational facility Beit Issie Shapiro, Raanana, Israel, all with independent mobility and with typical characteristics of RS stage III. The training took place at the educational facility, on a 1400 model treadmill (Trimline, capable of very low speeds < 0.5 k/h), with very long side rails. Special low side rails were adapted to the treadmill in order to fit the height of the children and velcro straps were added to assist in safely placing the hands. Pulse was monitored constantly during exercise by an A3 polar pulse belt. Pulse measurements at rest during training were considered as evaluators of aerobic physical condition. Functional measurement was based on a scale specially established for the present study. The scale was a 31-item motor-functioning tool that measures the ability of participants to knee walk and knee stand, to get up to a standing position, duration of walking different paths, and to go up and down stairs and slopes.

Results The study showed that physical fitness of the children at the end of the training programme had improved considerably ($P < 0.05$). Tests showed that general functional abilities had improved considerably ($P < 0.0001$). Although all items of the functional ability measure showed impressive positive change, some of the 31 items on it showed statistically significant improvement (knee walking, going up and down stairs and speed of walking for 25 m. Pearson correlation showed high linkage ($r = -0.76$) between functional improvement and change in physical fitness.

Conclusions Physical fitness programme executed on a daily basis is capable of improving functional ability of children with RS. Nonprofessional personnel can

Correspondence: Professor Joav Merrick MD, Medical director, Division for Mental Retardation, PO Box 1260, IL-91012 Jerusalem, Israel (e-mail: jmerrick@internet-zahav.net).

execute such a programme under supervision of a qualified physical therapist.

Keywords functional skills, physical fitness, Rett syndrome, treadmill

Introduction

The original study by Andreas Rett (1924–97) from Austria of two girls with a peculiar disease was published in German in 1966 (Rett 1966), but the syndrome only gained international attention, when Bengt Hagberg from Sweden and colleagues (Hagberg *et al.* 1983) published their findings, on 35 cases, in 1983.

Rett syndrome (RS) is a neurological disease characterized by arrest of brain development (Armstrong 1995) caused by an X chromosome mutation mainly affecting females (Amir *et al.* 2000). It is the first human disease found to be caused by defects in a protein involved in regulation of gene expression through its interaction with methylated DNA (Amir *et al.* 2000). It has been traced to a defective gene on the X chromosome called *MECP2* (pronounced meck-pea-two). This discovery was made by Huda Zoghbi, a neurogeneticist at Baylor College of Medicine in Texas working together with the Israeli physician Ruthie Amir, who also found the first mutations (Amir *et al.* 2000; Amir & Zoghbi 2000). RS typically presents after the first year of life (6–18 months of age), and is the most common cause for multiple-disability among females with an incidence of one in 10 000–15 000, with gradual reduction of speech and purposeful hand use, seizures, autistic like behaviour, ataxia, intermittent hyperventilation and stereotypic hand movements. After initial regression, the condition stabilizes and patients usually survive into adulthood (Hagberg 1985).

Although 50–75% achieve independent mobility at early childhood, about 75% loose the ability to walk at later years and become wheelchair bound (Lotan & Hadar-Frumer 2002). Some professionals recommend walking as a preventive intervention that might hold back or diminish secondary damage (McClure *et al.* 1998; Lotan 2000; Lotan & Hadar-Frumer 2002). One of the most debilitating disabilities is scoliosis, appearing in up to 85% (McClure *et al.* 1998). Poor prognosis for scoliosis has been linked with lack of walking and inability to climb stairs (Rossin 1997). Additional characteristic factors are constipation and osteoporosis at a young age; both situations are known to be positively affected by physical activity.

This population has been found to be at risk for developing sedentary lifestyle associated diseases, such as elevated risk for coronary artery disease, and stroke (DHHS 1996). Some researchers have suggested physical activity for children at risk in order to prevent old age health problems (Vaccaro & Mahon 1989). Such findings suggest the possibility that a physical activity programme might improve present and future health conditions.

There have been very few studies on physical exercise and RS (Larsson & Engerstrom 2001) and we have not been able to find studies on treadmill exercise with this population.

The purpose of the present study was to find out if activating children with RS on a treadmill, using a low intensity exercise on a regular basis, would improve physical fitness and/or functional ability with measurable results.

Methods

Study design

A few possible intervening factors were taken into consideration before executing the present study, including the initial functional ability of the participants, the stage of RS (there are four stages), the age of the child, other treatments, intensity of treatments and the experience of the therapist.

Because RS may manifest itself differently in each child, as a result of the limited number of cases on site and the fact that looking for other participants would add intervening factors, it was decided to use the research group, as its own control group.

Tests were performed at three intervals, designated times 1, 2 and 3, 2 months apart with intervention taking place between tests 2 and 3.

Participants

Participants of the study were four girls with RS aged 8.5–11 years (mean: 10 years) attending the educational facility Beit Issie Shapiro, Raanana, Israel, all with independent mobility and with typical characteristics of RS stage III. At this stage most individuals

with RS have difficulty performing purposeful hand movements, and motor skill decline and seizures are common. But there is actual improvement of behaviour, for example, less crying, irritability and less autistic-like behaviour. The child at this stage is more socially aware; attention span and communication efforts improve. Stage III can last for most of the affected person's life.

Procedures

The study was approved by each guardian and the physician in charge gave informed consent. Each participant was given two to four adjustment experiences on the treadmill at very low, personally adapted speeds (0.5–1.5 k/h) with monitoring at the 2-month period before intervention.

The training took place at the educational facility, on a 1400 model treadmill (Trimline, capable of very low speeds < 0.5 k/h), with very long side rails. Special low side rails were adapted to the treadmill in order to fit the height of the children and Velcro straps were added to assist in safely placing the hands. The pulse was monitored constantly during exercise by an A3 polar pulse belt.

Each child was trained on the treadmill while her hands were placed on the sidebars using Velcro straps and held hand-over-hand by the trainer. During training the songs preferred by each child (according to caregivers and parents) were played on a tape recorder. The daily training took place over a 2-month period. The primary level of difficulty (speed, duration of an exercise session and treadmill inclination) was individually adapted to the ability of each child, according to pre-intervention trials, using facial and body gestures as signs of fatigue and difficulty. The 2 months before intervention were used to observe and guide instruction by physiotherapist for trainer. Training was executed by a national service person (a person, who instead of military service, enrols in a programme of national service, such as working in hospitals, schools or special education), who was introduced to each participant, and individually instructed in regard to each participant, over a 2-week period, before the training sessions. Her role was to collect each child from her classroom (according to a careful schedule that ensured minimum disturbances to educational curriculum), to exercise the child according to the preset determinants and to write down pulse measurements for each participant.

Initial exercise sessions lasted 5 min, but were gradually lengthened over the first 3 weeks of training to 30 min each. The average session duration was later determined at 19.9 min. Each participant had 36–50 training sessions over a 2-month period averaging at 41 practice days for each child (the number of sessions depended on general health, and presence at school during the research period). The load of each training session (calculated by: speed × session duration × weight of the child × the sinus of the angle of inclination) ranged between 117 kc (Kilo Calories) and 245.7 kc and averaged at 213.5 kc. The overall work load taken by the participants during the course of the training period ranged from 3377 kc to 7243 kc (differences mainly because of weight differences between participants), averaging at 4599 kc, for each participant during a 2-month period. No weight loss was observed because of the training period, indicating that the children supplemented calories lost in training.

Measurements

Each test, at times 1, 2 and 3, included a physical fitness measure and a functional test. The physical fitness of the participating children could not be measured with formal tests because of the fact that such tests require walking to a distance of a mile (Fernhall *et al.* 1998) or half a mile (Rintala *et al.* 1997) and were beyond the capability of the participants. Because of the above-mentioned limitations, pulse measurement was used to evaluate aerobic physical condition at rest the lowest pulse during 5 min of seating, and during training, the highest measurement during 5 min of walking at 1.5 km/h, with 0 inclination. This method has been accepted in previous designs for this population (Tomporowski & Ellis 1985).

Functional measurement was based on a scale specially established for the present study. The scale was a 31-item motor-functioning tool that measures the ability of participants to knee walk and knee stand, to get up to a standing position, to walk at different speeds, and to go up and down stairs and slopes. The measurement tool took into consideration 12 levels of performance for each item recording the time it takes to complete a task, thus providing a consistent

grading from 0 to 372 points using seconds in time and meters in distance. The measurement tool was validated by a committee of three independent experts in child development and cognitive impairment. Inter-rater reliability for 20% of all measurements taken was found at a high level of 0.95. Reproducibility was checked between times 1 and 2 (a 2-month period with no intervention) and found at a level of 0.8. Execution of a single test can be performed through 30 min by an experienced examiner.

Results

All tests were analysed using a paired student *t*-test and Pearson *r* correlation.

Significant differences were found in pulse per minute and in motor functioning, when heart rate was measured at rest and during exercise on the three different time periods. The average heart rates at rest were found to be 111.0, 109.0 and 89.0 at times 1, 2 and 3 ($P < 0.05$ between times 2 and 3), while the average heart rates during activity were found to be 145.3, 145.0 and 121.5 at time 1, 2 and 3 ($P < 0.06$ between times 2 and 3). Despite the obvious difference in-between the findings before intervention and after intervention, and because of the small number of participants, differences were found only to have a tendency towards statistical significance ($P < 0.06$). In fact, the calculations revealed that in a case of six participants the differences detected were considered significant. The statistical differences in overall motor-functioning measurement between pre- and post-intervention were found to be highly significant ($P < 0.0001$).

In order to focus on specific findings, all 31 items were scanned and in each, a change was found between pre- and post-intervention measurement, at a level close to significant. However, four items were found to have a significant change between measurement taken before and after intervention, namely knee walking, going up and down stairs, and walking speed for a distance of 25 m.

For knee walking the difference in pre- to post-measurements ($P < 0.05$) resulted from the fact that three out of the four participants improved their knee walking ability. The reduction of time needed to pass 25 m by participants was found significant ($P < 0.05$), between pre- and post-intervention measurements because of two children improving their speed. Concerning descending a stair case, a most significant difference ($P > 0.001$) was found when comparing the pre- to post-measurements of the time it took the participants to go down a stair case of three stairs, because of the fact that all participants improved. Ascending a staircase, a significant improvement in the pre- to post-measurement ($P < 0.05$) was found in the ability of two of the participants to ascend a staircase without hand support.

If training improved both physical fitness and functional ability, there ought to be a correlation between the different findings. In order to detect that, a Pearson *r* correlation was performed between change in functional results pre- to post-intervention and the change in heart rate at rest pre- to post-intervention. The findings suggested a high negative correlation ($r = -0.76$), meaning that when heart rate goes down (improved aerobic condition), functional performance goes up.

A different way of examining the results would be to compare pre- to post-intervention functional advance. The results of such comparison showed that in the 2-month preceding intervention (spontaneous functional advancement) results were mixed. Two children slightly advanced, while the functional abilities of two children declined, resulting in an overall stability. On the other hand, when looking at the pre- to post-intervention period all participants showed considerable improvement.

Discussion

The present study is pioneering in its assumption that females with RS can be physically exercised with the use of a treadmill programme (at least with these four children at stage III, with some independent mobility). The study showed that a low intensity, daily training programme on a treadmill could reduce heart rates per minute, both at rest and during activity, which might be interpreted as improved physical fitness\aerobic capacity. Such results suggest, in this small sample, that basic physical fitness can be improved with a low intensity aerobic workout programme.

The study also found that children with RS can be introduced to a physical training programme per-

formed by nontrained persons after a short (2 weeks) individual training period, under close supervision of a physical therapist, thus reducing the cost of such an intervention and enabling the execution of a low-cost high-frequency programme. Such programmes can be performed in addition to physical therapy intervention and might enhance the results of such intervention. Results also showed that improved physical fitness might be linked with the improvement of functional abilities in the research population, thus enhancing the value of such programmes.

In two studies (Sekul *et al.* 1994; Ellaway *et al.* 1999), girls with RS had significantly longer corrected QT intervals and more T-wave abnormalities than age-matched healthy girls. With advancing stages of the syndrome, the proportion of corrected QT interval prolongations and T-wave changes increased. These findings suggest a possible cardiac basis for sudden, unexpected death in RS, which has resulted in parental caution against physical activity. Our findings on the other hand (again with four cases) showed that both physical fitness and functional abilities can be improved with a low-grade training programme, as long as the training intensity and duration of each session is gradually increased with close supervision over the trainee in regard to discontent and signs of distress, and as long as heart rate is constantly monitored.

One can question if the main motor improvement resulted from the treadmill training programme. When considering the overall improvement, we postulate that the common faculty to all areas of improvement (knee walking, accelerated speed in walking and ascending/descending staircase) is trunk control, or improved balance. This might explain why most items in the scale showed improvement. If we make such an assumption, then it is clear that treadmill training might be a good functional stabilizer for this research population.

Some of the findings carry further implications. For instance, the improvement in descending and ascending a staircase has been closely related to a better prognosis of scoliosis (Rossin 1997).

There are limitations to our study because of the small number of cases and we therefore suggest further research in the area of physical training of the RS population. The primary finding of the current research indicates that RS individuals show very low parameters of physical fitness. This coincides with the passivity that characterizes individuals with RS and to the fitness profile of people with developmental disability. Further investigation is warranted in physiologically profiling of the RS population, during strenuous activity under controlled circumstances, in order to design appropriate physical activity programmes that match their exact physical and physiological potential.

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