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CASE REPORT

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How reconstructive surgery combined with physiotherapy for a painful nontraumatic patellar dislocation enabled a woman with Rett syndrome to become pain free and remain physically active: A case report

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Key Clinical Message

The effects of orthopedic measures, with the exception of scoliosis surgery, are rarely described in individuals with Rett syndrome. In this case, treating a painful dislocation of the patella with combined orthopedic and physiotherapeutic measures enabled a woman with Rett syndrome to become pain free and remain physically active.

KEYWORDS

late motor deterioration stage, physiotherapeutic interventions, reconstructive orthopedic surgery, Rett syndrome

1 | CASE REPORT

Rett syndrome (RTT) is a rare neurodevelopmental disorder that affects approximately one in 10 000 newborn females.[1] The syndrome is characterized by an apparently normal psychomotor development during the first 6-18 months of life followed by a regression, accompanied by loss of motor skills and spoken words and a period of social withdrawal. A more stable period then occurs with the possibility to improve communication, fine motor, and gross motor skills again.[2] However, symptoms such as abnormal muscle tone, gait dysfunction, scoliosis, and stereotypic hand movements are commonly present. In adulthood, deterioration in motor skills may take place. The impairment profile of Rett syndrome has therefore been described as having four stages: early stagnation, rapid regression, stabilization, and late motor deterioration,[3] Table 1.

This case report was approved by the Ethics Committee in Umeå, Sweden, and written informed consent for participation and publication was obtained from the parent.

The purpose of the report is to highlight the importance of preventing a decline from the stable phase to the late motor deterioration stage with appropriate measures. The case describes a 39-year-old woman diagnosed with classic Rett syndrome. When she was a child, she rolled over at 5 months, sat without support at 6-7 months, crawled half-kneeling at 11 months, stood with support at 12 months, and walked independently at 19 months. She was left-handed and could hold a spoon but preferred to eat with her hands. Her eye contact was normal during the first 20 months; however, autistic features were suspected at the age of 22 months (the beginning of stage II). She started becoming less interested in social situations and became more unsettled. Hand stereotypies emerged, and she lost her hand function. Her posture became

TABLE 1 A short overview of the stage system for Rett syndrome

Early stagnation—Stage I This stage is characterized by an apparently normal psychomotor development during the first 6-18 mo of life where developmental milestones are achieved. This is followed by a period of stagnation
Rapid regression—Stage II Regression from the acquired developmental milestones is the significant characteristic of this stage. This is a period of social withdrawal, appearance of autistic features and loss of spoken words, fine and gross motor skills. Stereotypic hand movements emerge and additional medical problems such as seizures and respiratory abnormalities may become apparent. This stage can last between 1 and 4 y
Stationary stage—Stage III This stage is long and characterized by a relatively calm period where communication, fine motor, and gross motor skills can slowly improve again. Nevertheless, during this stage significant problems such as abnormal muscle tone, contractures, and scoliosis can emerge.
Late motor deterioration—Stage IV A decrease in, or lack of, mobility is characteristic of this stage. Some individuals may still be able to participate in a supported transfer while others constantly use a wheelchair. Problems can include worsening contractures and scoliosis. Seizures may be better-controlled and emotional contact improved

more flaccid. From time to time, she walked high up on her toes. At the age of two and a half years, the hand stereotypies had increased further. She began to use her hands again in feeding situations when three and a half years old and developed the ability to rise from sitting on a chair to standing, and back to sitting down again, to walk independently indoors and outdoors, and to walk up and down the stairs with gentle assistance (stage III). Her eye contact and social interest improved and her mood was generally calmer.

During the last years of her adolescence, a problem arose with lateral subluxation of her left patella in various situations, affecting her ability to flex her knee voluntarily. She began to walk with extended knees. Thus, independent walking became limited to when she was indoors and this activity was rarely practiced. Walking over doorsills became difficult, and walking on the treadmill was not feasible at all. During the transition from standing to sitting, she held her knee in an extended position, and thus, the position of her patella needed to be stabilized in a neutral position. This involved a carer using a firm handgrip around the patella to allow knee flexion (the beginning of stage IV). Her night sleep was greatly disturbed and the dislocation of her patella became increasingly painful. A knee orthosis was tested, but it did not manage to keep the patella in place.

As the intensity of the problems increased in her mid-thirties, the regional orthopedic team proposed surgery with the goal of the woman regaining her ability to walk and to reduce her pain. Since she was well known at the Swedish National Center for Rett syndrome and related disorders, a referral was sent to the orthopedic surgeon at the Center for a second opinion. The investigation at the Center showed that she had no spasticity, but had a contracture of her left adductor longus coupled with an increased valgus position and an extended left knee with an increased q-angle of >20°. A plain radiograph, including an axial view of the patella, together with an MRI of her left knee showed neither any trochlear dysplasia nor any femoral condylar dysplasia, nor insufficient ligamentous structures in her left knee. Upon gentle passive flexion to 40°, the patella dislocated and a severe pain reaction was observed. However, the patella dislocation could be reduced with a gentle extension maneuver of the joint. Four months later, the operation was carried out by the regional orthopedist.

During surgery, general anesthesia was used together with a peripheral nerve block through a catheter for intermittent administration of ropivacaine 2 mg/mL. During surgery, a tourniquet was applied for the left knee. The femoral nerve block was also used for 5 days for effective postoperative pain management. A combination of surgical procedures was carried out as follows: a proximal left tenotomy of the adductor longus muscle, release of the lateral patellar retinaculum together with a transfer of the semitendinosus tendon to the anterior aspect of the patella, distal and anterior advancement of the medial vastus of quadriceps, raphi of the medial patellar retinaculum, and a ventromedial transfer of the tibial tuberosity. A perioperative range of motion of 0-45° of flexion with a stable patellofemoral joint motion was achieved.

The femoral nerve block with an indwelling catheter was used for 5 days. A postoperative knee orthosis was applied. During the first 2 weeks, direct full weight bearing was allowed, but no flexion exercises were carried out. After 2 weeks, active and passive flexion was allowed from 0-30°. After 4 weeks, flexion between 0-60° was encouraged and after 6 weeks, no restrictions were implemented.

In collaboration with the regional orthopedic surgeon, targeted physiotherapeutic measures were introduced after the surgery. The main goal was for the woman to increase the degree of knee flexion and hip abduction and to regain the ability to walk again. The treatment from the physiotherapist started 2 weeks after the surgery and consisted of 5-10 repetitions of passive knee flexion and hip abduction, twice a week for the first 2 months after surgery, and once a week during the third and last months. Maximum passive knee flexion and hip abduction were performed with the woman in supine position and the hip at zero degrees of extension, and the knee was also flexed while she was in a sitting position. The maximum joint motion was kept for 30-60 seconds depending on the woman's mood or state of health. At the very beginning, the passive joint motions seemed to

make the woman uneasy, but they could still be performed by adapting the treatment. The duration of each session was approximately 30 minutes. In addition to the treatment from the physiotherapist, the woman's support network (parents and caregivers) performed 5-10 repetitions of passive joint motions of knee flexion and hip abduction four times a day and encouraged her to walk. The overall impression was that the treatment was tolerable for the woman based on her reactions, which were carefully monitored by the physiotherapist and the woman's carers.

Three months after surgery, an evaluation by the physiotherapist and a follow-up by the regional orthopedic surgeon were carried out. Both showed that the woman's knee flexion had increased from the initial 30° to 100° in a sitting position and to 80° in a supine position with the hip at zero degrees of extension. The hip abduction in a supine position with the hip at zero degrees of extension had reached 20°. The woman's ability to walk independently and on the treadmill was also regained. At the final follow-up by the regional orthopedic surgeon 6 months after surgery, the woman was pain free during night sleep, the passive joint motion and walking ability were maintained, and she could sit down on a chair and get up again.

The support network continued to encourage and assist the woman to use her motor skills in various physical activities. Four years after the surgery, the woman has retained the regained functions. According to her parents, she is mobile and can act independently, such as walking toward or away from an activity. She can actively flex her knee and sit down on a chair and get up again, walk on the treadmill and outdoors with gentle support. In addition to the gross motor improvements, she is also pain free, more present in social contact and, overall, seems happier.

2 | DISCUSSION

The orthopedic surgery, together with considerable tissue elongation measures and walking practice, prevented the decline from the stable phase to the late motor deterioration stage for the woman in this case. The exercises executed on a daily basis, assisted by the people within the woman's daily network, were crucial. The possibility to regain walking capability in adulthood after many years in a wheelchair has previously been raised in case reports.[4,5] More recent studies have also reported a more or less stable situation over time.[6-8] It is therefore vitally important to debate whether the late motor deterioration in the stage system for Rett syndrome should in fact remain or not. The stage might be taken as an unconditioned progressive phase of the syndrome leading to an expectation of motor deterioration in adulthood. Gross motor skills, for example walking, can diminish in any disorder due to secondary causes, such as orthopedic problems, poor health, and physical inactivity. As such, assessing body structures and functional motor skills, individual therapeutic programmes, and regular follow-ups are crucial throughout a patient's entire life. Unfortunately, regular physiotherapy is very seldom offered to adults with Rett syndrome by public health authorities. This lack of physiotherapy in adulthood almost certainly affects those persons' possibility to remain physically active.

The effects of orthopedic measures, with the exception of scoliosis surgery, are rarely described in individuals with Rett syndrome.[9] This is also true for the effects of physiotherapy for other orthopedic conditions than scoliosis.[5] This case contributes valuable knowledge in this regard. In conclusion, due to appropriate orthopedic and physiotherapeutic interventions, and with support from her network, the woman in this case regained pleasure in her life once she was able to be physically active and participate in daily events again without pain. As a result, a permanent decline to the late deterioration stage was prevented.

CONFLICT OF INTEREST

None declared.

AUTHOR CONTRIBUTION

LS: initiated the preparation of this case report for scientific publication and wrote the first draft of this paper. BH: provided all details about the surgery and post operation directions. PM: revised the paper critically and studied publications concerning orthopedic measures in Rett syndrome. All authors have reviewed and approved the submitted manuscript.

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