

## The Prosthetic Rehabilitation Of An Amputee With Rett Syndrome And Wrist Disarticulation

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### **ABSTRACT**

The rehabilitation process becomes very difficult, when a permanent disability as amputation is seen with a syndrome that requires special education as Rett Syndrome (RS). The importance of prosthetic fitting, and difficulties of rehabilitation in a 10 years old boy who underwent wrist disarticulation and having the diagnosis of RS is presented. The child couldn't be fitted with a prostheses at the age of 4 because of mental retardation and stereotypic movements. The patient who received special education for approximately 3 years, was given a long rehabilitation program following passive prosthetic fitting at the age of 7. Firstly, the subject had experienced difficulty in accepting the prostheses and presented aggressive behaviours. In time, he became aware of his loss and accepted the prostheses. It was thought that this situation in terms of showing the success of rehabilitation is important for the child amputee with a severe neurodevelopmental disease as RS.

**Key words:** Rett Syndrome, Amputation, Rehabilitation, Prosthesis

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### **INTRODUCTION**

Rett syndrome is a neurodevelopmental disorder that mainly affects females. It is characterized with cognitive disorder, communication problems, stereotypic movements and growth retardation (1,2). 1.09 of 10.000 girls up to 12 years have been reported as RS (1,3). It is the second leading disorder following Down Syndrome that causes severe intellectual dysfunctions in girls. MECP2 (methyl CpG-binding protein 2) gene mutation was detected in 90% of the individuals with RS (1,4).

Deterioration of motor functions and adaptive (cognitive, communicational) functions begins between 5-18. months. Motor function disorders like cognitive disorders, stereotypic hand movements, respiratory problems (hyperventilation), scoliosis and spasticity begin to appear (1,5). With this motor losses, a wheelchair is in the question for mobilization of the child (5).

Medications, surgical approaches and physiotherapy-rehabilitation programs usually take part in treatment of children with RS. These approaches aim at improving the symptoms and preventing the progression of the disease rather than treating the disease. In the context of rehabilitation program, education for daily life activities, various types of exercises to overcome physical problems and different physical therapy activities can be practiced (6,7). Walking education and utilization of supplementary technology products to facilitate walking and daily activities are also commonly applied approaches in RS (6). Speech therapy, education with music therapy are the other parts of rehabilitation (1, 2, 8).

Many factors that complicate the rehabilitation of subjects with RS are reported. Presence of problems that can lead loss of motor functions along with the symptoms of RS complicate the adaptation of the child to daily life and rehabilitation process. One of these problems that was not encountered in literature however that can lead this kind of problems in children with RS is amputation which is a permanent disability. Presence of amputations that could occur congenitally or acquired due to trauma, systemic disease in children with Rett syndrome along with motor function losses requires a comprehensive rehabilitation program and a multidisciplinary team approach.

Many cases of RS are available in literature (9, 10, 11, 12). However a case with RS and amputation was not found in literature. The study was carried out with the aim of determining the effect of prosthesis on rehabilitation of a male patient with RS who had amputation at the level of unilateral wrist disarticulation. The family was informed and an a permission for use of photographs was taken after informed consent was signed.

### **CASE REPORT**

The patient who is 10-year-old at present was amputated at the level of wrist disarticulation due to gangrene development following injection when he was 4. Stereotypic hand movements began in the subject who was diagnosed as RS at the time of amputation. Family of the patient of whom communication problems and dependence for self care gradually increased along with mental retardation applied to Department of Neurology for treatment and they were directed to Department of Physiotherapy and Rehabilitation in Faculty of Medical Sciences of H. U. in order to be informed about prosthesis.

When he applied to our unit, 4 months had passed over amputation, he was independent in terms of walking, sitting, standing up and remain standing however he was dependent to his mother for self care activities like eating, dressing and making his toilet. As the result of our observations, it was considered that making a prosthesis was not suitable for the patient who had stereotypic movements like taking his healthy hand and the stump to his mouth. His striving for putting the gloves off supported the opinion that he was not ready for using a prosthesis. The subject was followed-up by inviting for control at the same time with his appointment in Department of Neurology.



Family of the patient applied to our department again for prosthesis 3 years later. As the result of assessments, the patient who attended to a private education center was found not to have any physical and postural problems, stereotypic movements were found to decrease, muscle power was grossly found good, stump was unproblematic. It was observed that he had some difficulties for daily functional activities and dependence to family was observed to continue due to mental retardation despite good functional status. It was decided to make a passive wrist disarticulation prosthesis due to family's being willing to use prosthesis and in order to complete body image.

While process of taking the measurements was unproblematic, the patient's aspiration to take the socket out when stump-socket fit was being controlled caused this process to prolong. It was controlled whether there were any problems in the stump related to socket by sparking the child's interest to color, bright objects and toys and by not enforcing him. Other connections of the prosthesis were done after problems like wound, change of color were detected not to occur in the stump after a long trial period. In the trial period, the subject was observed not to accept the prosthesis again however not to take his part with prosthesis to his mouth. The patient who came to our unit every day for three weeks for prosthesis control and adaptation was noticed to get used to his prosthesis and not to try to take it out anymore. The family was informed about prosthesis, taking it on and off and advised to use it at home where he is more accustomed for short periods and control it after taken off, he was discharged with home program and invited for control 3 weeks later.

On his follow-ups at 3 week, 6 months and 1 year after discharge, he was observed to accept and notice the absence of the prosthesis. The family stated that the patient whose sockets were changed periodically and who is 10 years old at present still attended at the private education center, he also used his arm with prosthesis for support in activities requiring bilateral grasping and wanted to take his prosthesis on constantly, they did not experience any problems related to prosthesis however RS associated problems still continued.



## **DISCUSSION**

RS distresses the health team taking part in rehabilitation program and especially the family about overcoming symptoms and treatment. The condition becomes worse when a permanent disability like amputation accompanies it.

RS is generally seen in girls. However a few case reports in boys have also been reported (13,14,15). Our case is special also in terms of being a boy.

Rehabilitation of the 10-year-old boy with RS and who was amputated at level of wrist disarticulation with prosthesis required a quite hard and a long process. The facts that the family is aware of RS, being concerned with the child, being participated in rehabilitation program, the child's attending to a private education program for 3 years until prosthesis time are considered to have positive effects on making prosthesis and rehabilitation program. Otherwise prosthesis making process would be quite hard and may be even taking measurements could not be achieved.

Lacking of feed-backs due to mental retardation was one of the major disadvantages during stump-socket adjustment. Stump-socket adjustment, presence of any color changes in the stump related to socket or presence of a wound –in case of a such condition severe pain, rash and wound tissue development can come into question-was achieved by taking off and controlling the socket often. This process was kept longer than the stump-socket adjustment of the the patients with amputations at level of normal wrist disarticulation level in order to see whether aforementioned problems that could affect prosthesis use would develop. The family was informed and support was asked.

Active involvement of the amputee child is crucial in trial and utilization period when connections of prosthesis are done. Follow-up, control, taking the prosthesis on and off, playing games with prosthesis are in charge of mainly physiotherapist and family for amputee babies. Our case was regarded as a baby due to RS and mental retardation although he was 7 –year-

old and bilateral activities that could support the utilization of the prosthesis were chosen and tried to be done like playing games. In fact, expectation from the subject is only to accept the prosthesis, not to functionally use it.

One of the major problems in amputees is loss of body image after motor and sensorial losses and to provide the completeness of body image along with functionality expected by the amputee. When it was taken into consideration that our case is a child and an amputee with RS and the body completeness is crucial in RS patients, it is obvious that prosthesis use would be beneficial. Making prosthesis was initially begun for only this target however the prosthesis was adopted by the subject and was tried to be used in bilateral activities and culminated with his noticing its absence is gratifying in terms of the success of rehabilitation. This outcome is thought to contribute to rehabilitation team, to family and naturally to the child when such a case that is not available in literature is encountered.

### **CONCLUSION**

Rehabilitation of child amputees requires a hard and a long process. Rehabilitation of an amputee child with a neurodevelopmental disorder like RS is an intricate condition. However it can be easier and satisfying than expected with a good rehabilitation program, a prosthesis designed specially for and towards the requirements of the child and active participation of the child and the family in the rehabilitation program.

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