

Appendix 2. Other papers and literature important for clinical practice

Author	Participants	Title and Aim	Intervention/method	Outcome and Results
Baptista, P Mercadante, M Macedo, E Schwartzman, J (2006)	7	Cognitive performance in Rett syndrome girls using eye tracking technology	Study of eye movements and response to visual stimuli	Intentional gaze was measurable and it might be used to explore their cognitive performance
Bergström-Isacsson, M Larsson, G (2008)		The Musement music/motor function. DVD and book with therapeutic intentions, lyrics with chords and illustrative pictures	Using music, rhythm and movement in order to enhance motor activity and joy in children with Rett syndrome or any children	
Budden, S. S (1997)	60	Rett syndrome: habilitation and management reviewed	Clinical observation and experience	Hinged foot orthoses were useful for maintaining ambulation and a comprehensive team was important
Downs, J, et al (2009)		Guidelines for the clinical management of scoliosis in Rett syndrome	Consensus based on a panel of expert clinicians from Australia, Austria, Canada, Germany, Sweden, UK, USA Co-ordinated by J. Downs et al	
Downs, J Leonard, H Hill, K (2012)	12	Assessment of the StepWatch Activity Monitor™ to measure walking activity in Rett syndrome	A StepWatch Activity Monitor™ and video recording of activities were used	Persons with RTT were less active than healthy peers. There was reduced physical activity with age in spite of the ability to walk
Hanks, S (1990)	23	Motor disabilities in the Rett syndrome and physical therapy strategies	Clinical experience and medical records were used	Motor deviation was described and interventions suggested. No correlation between age of onset and severity of motor disability was found
Julu, P Witt Engerstrom, I Hansen, S Apartopoulos, F Engerstrom, B The ESSRA group (2012)	1	Treating hypoxia in a feeble breather with Rett syndrome	Assisted breathing using a positive airway pressure device in the CPAP mode	CPAP was successful in preventing tissue pO ₂ from falling below 49 mmHg. EEG amplitude during wakefulness improved during CPAP treatment

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Kadyan, V Clairmont, A. C George, R. J Johnson, E. W (2003)	1	Increased spasticity leading to functional decline in a woman with Rett syndrome	Intrathecal baclofen for spasticity management in Rett syndrome was used	A significant decrease in Ashworth scores was found, both upper and lower limb. Also improved range of motion, positioning, skin care, hygiene, and quality of life
Kerr, A. M Webb, P Prescott, R. J Milne, Y (2003)	91	Results of surgery for scoliosis in Rett syndrome	Clinical examinations and postal questionnaires were used for studying the severity of scoliosis and the effects of corrective surgery	Improved general well-being was found in 84%. Thirteen of 50 patients walked independently before surgery, and 12 patients did so after surgery; 2 stopped walking, and 1 who had not walked began to do so
Kjoerholt, K Salthammer, E (1990)	1	Intervention for Rett syndrome; with special reference to children and teenagers	Physiotherapeutic treatment, clinical experience	Suggestions of intervention and strategies related to personal experience
Larsson, E. L Aaro, S Ahlinder, P Normelli, H Tropp, H Oberg, B (2009)	23	Long-term follow-up of functioning after spinal surgery in patients with Rett syndrome	Evaluation of function after surgery of scoliosis was performed	Effect of surgery was: enough strength to keep the body upright, better seating position, less need for seating adaptations, reduction of time for resting during the day
Lotan, M (2011)		Rett syndrome: Physical therapy intervention	Book section in "Rett Syndrome: Therapeutic Interventions", Lotan, M and Merrick, J editors	
Prior, C Nunes, A Rios, M Sequeiros, J Maciel, P Gomes, L Temudo, T (2010) In Spanish with English abstract	25/7	Trastornos nutricionales y gastrointestinales en el síndrome de Rett: importancia de la intervención temprana. (Nutrition and gastrointestinal disorders in Rett syndrome: Importance of early intervention)	A previous protocol was used for assessing gastrointestinal function and nutritional status in RTT	Eleven out of 25 had feeding problems and individual intervention of 7 persons showed improvements in BMI, constipation and gastroesophageal reflux. Early individualized intervention is recommended

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Smeets, E. E Julu, P. O van Waardenburg, D Witt Engerstrom, I. Hansen, S Apartopoulos, F Curfs, L. M Schrander-Stumpel, C.T (2006)	1	Management of a severe forceful breather with Rett syndrome using carbogen	The NeuroScope system and EEG was used to monitor brainstem and cortical activity simultaneously in real-time. Carbogen by inhalation was used to prevent severe hypocapnoea attacks	There were reduced autonomic events, better night sleep and significantly improved quality of life
Wilfong, A.A Schultz, R.J (2006)	7	Vagus nerve stimulation for treatment of epilepsy in Rett syndrome	Adjunctive vagus nerve stimulation (VNS) therapy for a minimum of 12 months was used for persons with RTT	Seizure frequency was reduced at 12 months by $\geq 50\%$. Vagus nerve stimulation was safe and well tolerated with no surgical complications. For all there was increased alertness