

Reference for guidelines

Downs J , Bergman A, Carter P, Anderson A, Palmer GM, Roye DP, van Bosse H, Bebbington A, Larsson E, Smith BG, Baikie G, Fyfe S, Leonard H (2009). Guidelines for management of scoliosis in Rett syndrome patients based on expert consensus and clinical evidence, SPINE, Aug 1;34(17):E607-17.

The panel of expert clinicians

Gordon Baikie MD, Department of Developmental Medicine, Royal Children's Hospital, Melbourne, Australia

Allan C. Beebe MD, Nationwide Children's Hospital, Clinical Associate Professor, The Ohio State University, Assistant Orthopedic Program Director, Mount Carmel Health Systems, Columbus, Ohio, USA

Andy Bowe MD, Department of Orthopaedics, Robert Wood Johnson University Hospital, New Brunswick, USA

Allen Carl MD, Professor of Orthopedics and Pediatrics, Albany Medical College, Albany, New York, USA

Hilary Cass FRCPCH, Neurosciences Unit, Great Ormond Street Hospital for Children & Institute of Child Health, London

Eva Chan M Physio, Department of Physiotherapy, Glen Allan School, Melbourne, Australia

Craig P. Ebersson MD, Division of Pediatric Orthopaedics, Warren Alpert School of Medicine of Brown University, Hasbro Children's Hospital, Providence, USA

Carolyn Ellaway PhD, Western Sydney Genetics Program, Children's Hospital at Westmead and Discipline of Paediatrics and Child Health, University of Sydney

Peter Gibson FANZCA, Department of Anaesthesia, Children's Hospital at Westmead , Sydney, Australia

Jocelyn Goodall PT, Pediatric Development and Rehabilitation Department, Legacy Emanuel Children's Hospital, Portland, Oregon, USA

Robert N. Hensinger MD, Department of Orthopaedic Surgery, University of Michigan, Ann Arbor, Michigan, USA

Peter Huppke MD, Department of Paediatrics, Georg-August University, Gottingen, Germany

Walter Kaufmann MD, Centre for Genetic Disorders of Cognition & Behavior, Kennedy Krieger Institute, School of Medicine, Johns Hopkins University, Baltimore, USA

John Killian MD, Orthopaedic Specialists Alabama, Birmingham, USA

Alexander Krebs MD, Orthopaedic Hospital Vienna Speising, Department of Children Orthopaedics, Vienna, Austria

Eva-Lena Larsson PhD, Orthopaedic Centre, University Hospital, Linköping, Sweden

Gunilla Larsson MSc, Swedish Rett Centre, Frosön, Sweden

Bruce McPhee FRACS, Chair of Orthopaedic Surgery, University of Queensland, Brisbane, Australia

Ulrich Meergans MD, Department of Orthopaedics, Seepark Klinik Langen-Debstedt, Germany

Peter Newton MD, Pediatric Orthopaedic and Scoliosis Center, Rady Children's Hospital, San Diego, USA

Roy Nuzzo MD, Department of Orthopaedics, Overlook Hospital, Summit, USA

Greta Palmer FANZCA, Department of Anaesthesia and Pain Management, Royal Children's Hospital, Melbourne, Australia

Alan K. Percy MD, Civitan International Research Center, University of Alabama, Birmingham, USA

James Policy MD, Department of Pediatric Orthopedic Surgery, Director of Spinal Deformity, Children's Hospital Oakland.

David Roye MD, Division of Pediatric Orthopaedic Surgery, Morgan Stanley Children's Hospital of New York Presbyterian, Columbia University Medical Center, New York, USA

Suken A. Shah MD, Department of Orthopaedics, Alfred I. DuPont Hospital for Children, Nemours Children's Clinic, Wilmington, Delaware, USA

Brian G. Smith MD, Department of Orthopedics, Yale University School of Medicine, Yale New Haven Children's Hospital, USA

Ian Torode FRACS, Department of Orthopaedics, Royal Children's Hospital, Melbourne, Australia

Stephen Tredwell MD, Professor Emeritus Department of Orthopaedics, University of British Columbia, Canada

Hans Tropp PhD, Department of Orthopaedic Surgery, University Hospital, Linköping, Sweden

Harold van Bosse, MD, Department of Orthopaedics, Shriner's Hospital for Children, Philadelphia, USA

Lyn Weekes FCSP, Rett Syndrome Association-UK and Rett Syndrome Association Scotland, UK

Alexander Wild MD, Department of Orthopaedics, Hessing Stiftung Augsburg, Germany

Matthew Yates BPTy, Cerebral Palsy Education Centre Inc., Melbourne, Australia

Parents provided feedback on the clinical areas covered in the guidelines.

Development of guidelines co-ordinated by Jenny Downs PhD, Anke Bergman MPH, Philippa Carter MBBS, Alison Anderson BSc (Hons) , Ami Bebbington BSc (Hons), Sue Fyfe PhD and Helen Leonard MBChB at the Telethon Institute for Child Health Research, University of Western Australia, Perth Western Australia.

Guidelines for clinical management of scoliosis in Rett syndrome

Why we developed the guidelines

Scoliosis is a common orthopedic complication of Rett syndrome with about three quarters of girls affected by the age of 13 years. Families have indicated the need for more information on scoliosis management and the topic of scoliosis is also often discussed. There is limited peer-reviewed literature to guide clinical management although there are many experienced clinicians in different countries. Therefore, the aim was to develop guidelines for the clinical management of scoliosis in Rett syndrome by pooling information gained from review of the literature and the opinions of expert clinicians.

How we developed the guidelines

The first draft of the guidelines was written after a comprehensive review of the literature and contained a list of potential statements describing practice and questions for where the literature was lacking or unclear. We also asked a panel of parents of a child with Rett syndrome to check that the content of this draft addressed their concerns. An international panel of expert clinicians from different specialties then reviewed successive drafts until agreement was reached.

The guidelines

The specific guidelines are listed on the inside pages, and they identify the main aspects of clinical care relating to the health of girls and women with Rett syndrome and scoliosis. They are not a recipe for the management of all cases but rather give broad guidance. They have the potential to support clinicians with less experience, promote discussion between clinicians and parents, and to stimulate research to improve the evidence for management.

Full details of the paper describing the development of the guidelines are available in the reference shown on the last page of this leaflet. The members of the panel of expert clinicians are also listed on the last page and we acknowledge their contributions.

To accompany this leaflet, we have developed a booklet that further describes the guidelines and includes relevant quotes and photos from families with a daughter with Rett syndrome and scoliosis. Please talk with your doctor if you have questions about the information contained in this leaflet or the booklet.

The Rett Syndrome Association of Australia and the International Rett Syndrome Foundation have contributed to the funding of the production of this leaflet.

Guidelines for the clinical management of scoliosis in Rett syndrome

1. Monitoring and intervention in regard to the diagnosis of scoliosis

- 1.1 Physical assessment of the spine should be conducted at the time of diagnosis of Rett syndrome and at least every 6 months thereafter.
- 1.2 Because of the high prevalence of scoliosis in Rett syndrome, families should be given information about this early in the child's clinical course.
- 1.3 All children with a clinical diagnosis of Rett syndrome should undergo genetic testing as genotype may influence the development and management of scoliosis.
- 1.4 Therapy should aim to:
 - Develop, maintain and promote walking for as long as possible.
 - Optimize strength of back extensors.
 - Maintain flexibility of the spine.
 - Implement a postural management scheme that includes appropriate support for correct sitting posture and sleeping posture supports.

2. Monitoring following a diagnosis of scoliosis

- 2.1 Referral should be made to an orthopedic surgeon when there is clinical concern regarding scoliosis
- 2.2 Physical examination of the spine should be conducted at least every 6 months, but the orthopedic surgeon may increase the frequency of assessment in the following situations:
 - Abnormal early development/never learning to walk.
 - Low muscle tone.
 - During growth spurts.
 - Early age of onset.
 - Greater Cobb angle.
 - Children with genotypes known to be at higher risk of more severe scoliosis (p.R168X, p.R255, p.R270X).
- 2.3 Physical assessment in Rett syndrome should include:
 - Sitting balance and symmetry of weight bearing in sitting.
 - Level of walking ability and time spent walking.
 - Total distance walked.
- 2.4 At each visit, height and weight should be measured.

3. Imaging

- 3.1 Request an initial X-ray if there is evidence of a curve.
- 3.2 It is preferable to assess skeletal maturity with a hand and wrist radiograph but assessment of the iliac crest growth plate is also an option.
- 3.3 Six monthly X-rays are suggested if the Cobb angle is more than 25 degrees before skeletal maturity and twelve monthly X-rays after skeletal maturity until evidence of no further progression.
- 3.4 Plain radiography is sufficient in monitoring the progression of the curve. The following views should be obtained and should include shoulder to pelvis:
 - Standing upright AP and lateral spinal radiographs for patients who can stand at their initial visit.
 - Sitting AP and lateral spinal radiographs for patients who cannot stand.
 - Supine AP and lateral spinal radiographs for patients who cannot sit.
- 3.5 AP films alone may be used in follow up x-rays.

4. Therapy and conservative management

- 4.1 Involve physiotherapists and occupational therapists as soon as scoliosis has been diagnosed.
- 4.2 Physiotherapy should be used to maintain musculoskeletal well-being in children with Rett syndrome and scoliosis. There is not yet evidence that physiotherapy will prevent progression of an established scoliosis.
- 4.3 Aim to prolong ambulation as long as possible. Aim to increase the distance that the child can walk and/or the length of time the child can stay on their feet (at least 2 hours per day where possible).
- 4.4 For those who cannot walk, use standing frames for at least 30 minutes a day.
- 4.5 Aim to maintain range of movement of joints.
- 4.6 Symmetrical seating is valuable for the child's comfort and functioning.
- 4.7 Assess, monitor and optimize Vitamin D levels. Improve dietary intake of calcium and time spent in daylight to promote bone health.
- 4.8 In severe scoliosis where the risks of surgery outweigh the benefits, the management plan should include:
 - The provision of supported seating to optimize posture.
 - Monitoring and treatment of pressure sores.
 - A low threshold for antibiotic use during respiratory infections to minimize the effects of restrictive lung disease.

5. Spinal Bracing

- 5.1 There is no consensus that bracing is beneficial in reducing the progression of scoliosis in Rett syndrome but it may be used if seating and trunk activation cannot be achieved.
- 5.2 If tolerated, bracing should be used in the skeletally immature child, to help delay surgery.
- 5.3 The following potential complications of bracing must be considered: pressure sores, respiratory impairment, discomfort, skin irritation, exacerbation of gastro-oesophageal reflux and the potential to decrease trunk strength, flexibility and physical activity.

6. Pre-operative considerations

- 6.1 Surgery should be performed in a specialist center due to the high risk of anaesthetic and post-surgical complications.
- 6.2 Surgery should not be delayed until skeletal maturity, however, caution should be used before performing surgery in children younger than 10 years of age due to the following problems: decreased trunk height, pulmonary restriction, 'crankshaft', and secondary curvatures.
- 6.3 Surgery should be considered when the Cobb angle is approximately 40 to 50 degrees.
- 6.4 Surgical objectives should include:
 - Achieving a spine that is balanced and fused.
 - Restoration of the normal sagittal profile.
 - Achieving level shoulders and hips.
 - Improving the well-being and functioning of the child.
 - Improving carer well-being.
- 6.5 There should be a period of hyperalimentation if weight is less than the 5th centile.
- 6.6 The following markers of nutrition should be assessed: BMI, Hemoglobin, electrolytes, albumin (<3.5mg/dl), white cell count.
- 6.7 Patients with Rett syndrome need special anaesthetic consideration in line with other neuromuscular disorders. They are highly sensitive to analgesia, sedatives and volatile anaesthetic agents.
- 6.8 In addition to the pre-operative assessment used in all scoliosis surgery, the following must be considered before anaesthetizing a child with Rett syndrome:
 - Breathing patterns (hyperventilation, breath holding).
 - Pre-operative arterial blood gases/capillary gases.
 - Gastro-oesophageal reflux.
 - Autonomic disturbance.
 - Seizure history, management and medications.
 - Pre-operative ECG to identify possible prolonged QT syndrome.

7. Surgical considerations

- 7.1 In the majority of cases it will be possible to use a posterior-only spinal fusion. This is the definitive management of neuromuscular scoliosis in girls with Rett syndrome.
- 7.2 If anteroposterior surgery must be used, a single-stage approach is preferable in order to reduce anesthetic and surgical complications but a staged procedure may be appropriate in the presence of significant co-morbidities.
- 7.3 Fixation to the pelvis may be indicated if pelvic obliquity exists in the non-ambulant child. There is no consensus about the degree of obliquity that indicates fixation.
- 7.4 If a reliable signal can be obtained, Motor Evoked Potentials and/or Somatosensory Evoked Potentials can be used to detect neurological injury during neuromuscular scoliosis surgery.

8. Post-operative considerations

- 8.1 Admit to HDU/ICU post-operatively.
- 8.2 Care needs to be taken with regards to the titration of analgesia so that pain relief is adequate and sedation is minimized to ensure respiratory effort is not compromised. Post-operative analgesia must be closely monitored by a specialist pediatric pain team with 24hr cover or intensive care specialists.
- 8.3 Frequent and aggressive chest physiotherapy should be used. Non-invasive positive airway pressure support may be required post-extubation (e.g. BIPAP)
- 8.4 A clear management plan should be constructed when the patient is transferred back to the ward.
- 8.5 Seek expert advice to optimize nutritional status.
- 8.6 Consult parents or caregivers to help assess the child post-operatively.
- 8.7 Mobility post operatively:
 - Log roll for bed mobility.
 - Sitting on edge of bed day one post-op.
 - Transfer to chair post op day two.
 - Walking (if possible) post op day three.
- 8.8 Post-operative reviews should be carried out at:
 - 6 weeks.
 - Then every two-three months over the first year.
 - Annually thereafter.
- 8.9 The following should be used to assess surgical outcome: complications including bleeding, infection and duration of ICU stay; Cobb angle and achievement of fusion; respiratory status; sitting balance, function and quality of life; parent and carer satisfaction.