

International Guidelines for Management of Communication in Rett Syndrome.

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Background:

Rett syndrome (RTT) is a rare disease caused primarily by de novo mutations in the X-linked *methyl-Cp2G-binding protein 2 (MECP2)* gene.

- It affects around 1:9,000 females and is associated with language & physical impairments, which contribute to severe difficulties with communication.
- Being able to communicate allows an individual to express needs & wants, make choices, offer opinions, build relationships, access education, and participate in society.
- Difficulties in communication have a profound impact on quality of life.
- Caregivers report their struggle to access services & support tailored to an individual's specific communication needs.
- There is wide variation within & between countries.

Aims of the project:

Two-year project to improve quality of life for individuals with RTT, by producing international guidelines that will:

- Provide a basic common framework for good practice.
- Be adaptable to services & individuals across countries.
- Be available online & through national and international Rett organizations and parent associations.

Conclusion:

- Fundamental to the effective uptake of the guidelines is the involvement of individuals with Rett syndrome, their families and professionals who work with them. The project draws on knowledge & experience from as many countries as possible. It is firmly embedded in the lived experience of Rett syndrome.
- Strategies for dissemination, training & implementation include webinars/teleconferences, spin-off projects with parent associations, & utilisation of professional support networks.

The guidelines are due to be published at the end of May 2018.



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Methods:

Led by an international consortium, the project has combined 'available evidence' with 'expert consensus', by:

- Conducting systematic reviews of published & grey literature.
- Taking an inventory of clinical practices around the world.
- Consulting with parents/caregivers and professionals through focus groups & online surveys (available in 16 languages).
- Extracting themes and statements from the reviews, inventory & surveys to create draft guidelines.
- Applying a 2-stage (Delphi) review process with support from an "expert" group to finalise the guidelines.

Results:

- 250 statements were reviewed in Delphi Round One and 101 in Round Two.
- The majority of statements exceeded a pre-set consensus level of 70%.

Final guidelines = 271 statements/recommendations:

- Guiding principles (rights and beliefs) = 20.
- Professional Practice (principles of teamwork, responsibilities of professionals, Rett specialist clinics and expertise centers) = 29
- Features of Rett syndrome that impact communication = 19
- General strategies for engaging with individuals with Rett syndrome = 26
- Assessment of general communication skills (including principles, people & context, procedures & settings) = 42
- AAC assessment (including readiness for AAC, models of best practice, components of assessment) = 23
- Assessment through trials of AAC system/device = 33
- Intervention (including principles, target setting, tools & techniques) = 21
- Intervention goals = 58

The guidelines are underpinned by the *UN Convention on the Rights of Persons with Disabilities* & by the fundamental belief that **"All communication partners should believe that the individual with Rett syndrome is capable of communicating"**.

