

10.6 The ICF and Rett Syndrome

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Introduction

Rett syndrome is a relatively rare but serious brain disorder most often affecting girls. Generally, the early development of an infant with Rett syndrome appears normal. However, some time in the first 6 to 18 months of life, the normal pattern of childhood development does not continue (Kerr & Witt-Engerstrom 2001).

In most cases there is a loss of communication and hand skills with the subsequent development of unusual hand movements. In the long term, most children are no longer able to talk, and many find it difficult or are unable to walk. Other clinical features include poor head growth, epileptic fits, spinal curvature, abnormal breathing patterns, and gastrointestinal and sleeping problems. Rett syndrome is, therefore, usually associated with severe intellectual and physical disability, and girls with this condition commonly suffer from considerable health problems. Despite this, however, many parents remark that over time girls may appear to improve in their social awareness and communication skills.

Rett syndrome is a genetic disorder and has recently been shown to be associated with mutations in the methyl-CpG-binding protein 2 (*MECP2*) (Amir et al. 1999). The *MECP2* gene is on the X chromosome. However, despite these recent genetic advances, there is still little information available about the natural history of the disorder, and we do not know which factors (in the girl, her family and her broader environment) are related to prognosis.

The principal aim of the Australian Rett syndrome research program, which began in 1993, was to define a population based cohort, that could be used for subsequent clinical and epidemiological studies and followed prospectively (Leonard et al. 1997). Baseline data on communication, mobility, symptoms and classification have been gathered since 1993 on individuals as they are enrolled in the cohort. In 2000, data were collected on functional ability in daily living, behaviour, hand function, medical conditions and use of health, therapy and education services (Colvin et al. 2003). In addition, parents used a calendar system to report the occurrence of medical, other health and therapy appointments for the duration of that year. Molecular testing for the presence of *MECP2* mutations has also now been undertaken on more than 80% of cases, with mutations identified in approximately 70% of cases.

The research program has several aims:

- to assess the burden of Rett syndrome on the affected individual, their family and the community
- to determine which factors contribute most to this burden and which factors may alleviate this burden by determining:

- how functional ability, behavioural score and medical/health burden at specific ages is influenced by a range of environmental and genetic factors
- how the outcome for the family, in terms of quality of life, is influenced by the child’s genetic characteristics, family functioning, support and resources.

Thus the components of the ICF—

- Body Functions and Structures
- Activities
- Participation
- and contextual factors (Personal and Environmental Factors)

provide a conceptual framework for investigating the broadest aspects of disability and functioning in Rett syndrome.

Body functions and structures

A number of body functions and structures studied in the Rett syndrome research program are relevant to the ICF:

- mental functions/nervous system
- sensory functions/eye, ear
- voice and speech functions
- functions of digestive, metabolic, and endocrine systems
- genitourinary/reproductive functions
- neuromusculoskeletal and movement-related functions
- functions of the skin and related structures.

The ICF framework provides the capacity to identify variation in body function and structure.

In our long-term follow-up studies, we specifically inquire in our parental questionnaire about medical conditions associated with Rett syndrome (e.g. scoliosis, digestive disorders, and epilepsy). During our 1-year intensive calendar study, parents reported on their daughters’ health on a daily basis, which allowed us to gain a window into the day-to-day life of a girl with Rett syndrome.

The assessment of intellectual function is particularly difficult to assess in Rett syndrome because of the apraxia and communication difficulties associated with this disorder. However, we have used the WeeFIM (as is described subsequently in the activities and participation sections) to obtain an estimate of cognitive functional ability (Leonard et al. 2001). Neurological functioning is of particular interest as two-thirds of girls with Rett syndrome suffer from epilepsy (Glaze et al. 1987). In our study, parents document the occurrence of epileptic seizures, which will allow us to categorise their seizure profile.

Visual and hearing function have not been specifically assessed in our research program as these are thought to be normal in Rett syndrome. However, autonomic functioning (in particular involving respiratory and cardiovascular systems) is of interest as it is thought that there is a neurologically mediated disturbance in these areas (Julu et al. 2001). Parents describe the nature of breathing disorders (e.g. hyperventilating) and patterns of occurrence. This has the potential to impact on their growth as it is thought that the 'work' of hyperventilation (or deep breathing) is associated with high levels of energy expenditure (Motil et al. 1994). In addition, clinical studies carried out by our colleagues at the Children's Hospital at Westmead (Sydney) will involve the use of the 'Autonomic Monitor' to specifically test this function.

The investigation of digestive system functioning is relevant to Rett syndrome because girls with this condition are commonly affected by gastro-oesophageal reflux disease and constipation as well as growth problems (Motil et al. 1999). Our questionnaires include detailed questions about feeding patterns including the quantity of meals and the time taken to feed. As well as assessing digestive system functioning, we also attempt to assess the way in which these functional problems can be overcome to optimise the subject's wellbeing as well as minimise the burden on families and carers. One example is the use of PEG (percutaneous endogastric tube) insertion to increase caloric intake, decrease feeding time and improve weight and overall health.

The presence of scoliosis, which may be treated by physiotherapy, bracing and/or surgery, is an example of abnormal body structure that affects up to half of girls with Rett syndrome (Stockland et al. 1993). We are currently undertaking a study to assess the impact of these treatments on the progression of scoliosis and on the functioning of girls and women with Rett syndrome. In addition, structural impairments affecting mobility can be overcome with the use of aids such as wheelchairs and walking frames. We are attempting to quantify the use of these aids and identify any problems or restrictions to their access.

The structural and functional deficits associated with Rett syndrome often require hospitalisations for the purpose of managing acute medical conditions as well as operative procedures to correct problems. These hospitalisations result in a burden on the health system as well as the families who are caring for their children during the illnesses. We are attempting to quantify the impact that this population has on the health system, in terms of hospitalisations, medical visits and allied health services, by analysing data collected from the 'daily calendar' is completed by parents.

Finally, the genetic make-up of an individual can be considered to be part of body structure. As described above, our epidemiological study also includes the mutation status of each individual and the extent to which they have skewing of the X chromosome.

Activities

Our questionnaires include sections about the individual's activities in a number of domains relevant to the ICF:

- activities of learning and applying knowledge
- communication activities
- movement activities
- self-care activities
- domestic activities
- interpersonal activities
- performing tasks and major life activities.

The subject's ability to perform practical, day-to-day communication activities is assessed using the WeeFIM. This is a functional independence measure that we have used to measure independence in activities of daily living taking into account the help and assistance that may be required. Items include the extent of comprehension and ability to verbalise. We also have information about the subject's ability to communicate at all stages of her life, e.g. before she went through regression. Parents are asked to describe equipment used to aid communication.

Mobility is also formally assessed using the WeeFIM, which covers locomotion, toilet/bath and shower transfers and chair/wheelchair transfers. Ability to manipulate objects with the hands is an item of interest in our research because one of the hallmark features of this disorder is loss of functional hand use. We use the hand apraxia score to attribute a value to the degree of functional hand use (Burd et al. 1990). Furthermore, the use of hand preference in Rett syndrome has also been studied in depth as a result of information collected from parents (Umansky et al. in press).

Activities of self-care are again assessed using the WeeFIM. Specific questions are devoted to bathing, dressing, grooming and toileting. Parents document their daughter's ability in this domain and, as well as scoring her on a predetermined scale, they include extra written information about how she carries out these activities. We include questions about puberty and menstruation in our long-term study to assess sexual development in this population, as this has implications for self-care and parental management. As already indicated, feeding and meal times are important elements of our research because of the practical implications to the child and family. Parents document the quantity of food provided, preparation procedures and mealtime routines. Information about specific equipment used with meals to facilitate feeding and oral functioning is also requested.

Girls and women with Rett syndrome are obviously unable to take an active role in traditional domestic activities such as shopping, meal preparation and housework. However, parents and carers may make an effort to include the girls and women where possible in these activities, e.g. by including them in shopping trips and having them in the kitchen during meal preparation so that they can feel part of the

activity. We recognise the potential therapeutic benefit of these activities and parents are therefore able to document them in a section on day-to-day therapy.

In some cases, communication, movement and interpersonal activities have been assessed using a video protocol, whereby families and therapists record the subject performing, where possible, a number of structured tasks. These procedures are preferably conducted in a familiar setting, e.g. home or school to maximise functional ability.

Participation

Our questionnaires include sections about the child's or young adult's participation in a number of domains relevant to the ICF:

- personal maintenance (i.e. self-care)
- mobility
- exchange of information
- social relationships
- home life and assistance to others
- education
- community, social and civil life.

By the nature of their disability, girls and women with this condition are not usually able to participate in work, employment and economic life in the traditional manner.

The extent to which subjects can take part in their personal maintenance is assessed using the self-care domain in the WeeFIM. In addition, parents are able to give quantitative answers about their daughter's ability to care for herself. In general, a subject is usually limited in her ability to participate in personal maintenance but may be able to ease the burden on caregivers, e.g. by indicating when she is soiled. Participation in nutrition is usually limited because of the severity of the disorder, but parents are able to document choice making at meal times, e.g. through use of photographic cards or actual foods. Taking part in a range of therapy services (e.g. physiotherapy, occupational therapy, music therapy, hydrotherapy) can be considered to be participation in health. Using the calendar system, parents documented participation in health services over a period of 1 year during 2000. The extent to which subjects take part in therapy may be influenced by many factors. In our studies, we have found that therapy participation varies according to severity of disability, age and maternal education (Moore 2002).

Mobility is an important area of functioning in girls and women with Rett syndrome because it is often compromised, resulting in a decreased ability to participate in the community (Kerr & Witt-Engerstrom 2001). We have collected specific information about subjects' abilities to mobilise, whether they can do so independently, or by being supported or fully aided with equipment. Mobility is of central importance, and information has been sought about parent's attempts to modify the home environment and their vehicles for this purpose.

Participation in exchange of information can be difficult to assess. Again, the WeeFIM covers social involvement by quantifying the degree of assistance the girls and women require to express their needs and feelings. In addition, the use of communication devices in Rett syndrome is documented in the follow-up study. As well as objectively quantifying the degree of functionality in this domain, parents are able to describe the way in which their daughter communicates e.g. facial movements, gestures. This is closely related to the development of social relationships and ultimately to participation in community, social and civil life. The challenge is to find ways of communicating that will maximise the involvement of subjects and their families. The relationship between the girl/woman and her siblings is explored.

Participation in home life and assistance to others is obviously related to the degree of disability in Rett syndrome. Respondents indicate the accommodation–living situation in the questionnaire. Increasingly, young women with Rett syndrome are gaining access to group homes and supported living with the assistance of government organisations. This is allowing these women to take on a role in a home environment with like-minded peers.

Participation in education by girls and women with Rett syndrome has undergone significant change during the past 10 years in keeping with shifts in philosophy and policy in special education. In our follow-up studies, parents have been documenting the educational setting as well as the amount of aide support for their daughters. Parents are asked to provide further information about their degree of satisfaction with their child’s school arrangements. Furthermore, the participation in informal educational activities (e.g. in the home setting) is included.

Participation in community, social and civic life is an important element of life for children and women with Rett syndrome. Parents have documented participation in family events, religious events such as First Holy Communion, school events such as sports carnivals, and ethnic celebrations such as St Patrick’s Day. Participation in recreation and leisure activities will also be specifically included in the upcoming parental questionnaire. This will include activities such as swimming, horseriding and attending cinemas.

Participation facilitation can be related to the extent to which therapy and education professionals are informed about Rett syndrome. Participation restriction is also likely to be related to a number of factors. These include the physical disability and immobility associated with Rett syndrome, which impede access to events/activities. In addition, participation in life situations can be somewhat impaired because of communicative disabilities associated with the disorder and parents’ lack of resources for overcoming this aspect of the disability (i.e. communication devices). The attitudes of service providers are also important.

The Australian Rett syndrome research program is based on a biopsychosocial model which integrates aspects of both medical and social models of disability and functioning. The investigation of environmental factors such as equipment and support available to individuals and families and the social capital of the

communities in which they live is likely to be integral to understanding the burden of this disorder. The program will use the ICF framework to identify those factors determined to be most beneficial and cost-effective in optimising health, function and quality of life for the affected child and her family.