

The services to persons with Rett Syndrome in Norway and the Frambu model empowering users and families

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Rett Syndrome in Norway

- 1983: The first girl diagnosed with RS in Norway.
- 1987: The Norwegian Rett Syndrome Association (NRSA) was founded during a family course at the Frambu Resource Centre for rare Disorders.
- 2014: NRSA has about 120 members diagnosed with RS. In addition there are about 30 more persons with RS, totaling 150 persons with RS in Norway.
- A research project on different aspects of RS in Norway is currently being planned.

What is rare?

- When less than 100 persons per 1 million inhabitants (i.e. 1: 10 000) have the same condition/syndrome/disorder/disease, it is defined as rare.
- In Norway with 5 million inhabitants this means less than 500 persons are affected by the same thing.
- It is estimated that there are 30 000 people living with a rare disease in Norway, compared to 30 million in Europe (less strict definition).

Services for persons with Rett Syndrome in Norway

- Assessment and diagnosis by the ordinary health care system
- Genetic assessment and counselling during or after the process of giving the diagnose.
- Follow-up by
 - the family doctor in the community,
 - the child specialist at the county pediatric department/OPD or habilitation unit,
 - Or generally specialized, though not syndrome specific clinics, in regional or university hospitals
 - Other medical specialists according to needs/symptoms,
 - When passing 18 years of age: Follow-up by habilitation unit for adults, and medical specialists according to needs

Services for persons with RS in Norway (cont.):

- Health and dental care services are free for children <18 y; thereafter health care is essentially free after exceeding the general "annual fee" of about 200 Euros.
- A locally based **responsibility group** around the person with RS and her family is established in the community, consisting of local professionals as needed.
- It meets regularly (2-4 times/year) to discuss challenges, sort out problems and areas of concern and take actions accordingly, refer to specialists, investigations etc.
- The group is also responsible for making an **individual plan** (IP) for the present/coming time as well as for the future 2-5 (-10) years, in areas like needs for relief or assistance, auxiliary housing, respite care, sheltered home etc.

At the community level there will usually be services and follow-up by:

- Responsibility group
- Family doctor
- Health station and community health nurse
- Physiotherapist
- Ergonomist/occupational therapist
- Specially trained teacher/educational therapist
- Communication therapist
- Psychologist
- Community dentist
- Etc.

In Norway there is

- No Rett centre or Rett clinic that only deals with RS
- **Frambu Centre for Rare Disorders**, that offers different kinds of services for persons with RS, their families and the network of professionals around them. This is because RS is a rare disorder, allocated to Frambu as one of 120 different rare disorders, sharing the same centre.
- Frambu acts as **an advisory unit** for families and professionals searching for information and experience. Anybody may contact a professional at Frambu, by phone or mail, and discuss, ask, get advice etc.
- Frambu has produced **lots of information**, free and available for everybody, in Norwegian, but Google translate will do the translation to (almost) any language!



Frambu













Short history of Frambu

- Established and built by voluntary work by the youth organization of the Norwegian labour movement in 1954, inaugurated in 1955.
- Situated in the forest 17 km south of Oslo city centre, but sharing boundaries with the capital in the middle of the lake!
- In the beginning the centre arranged courses for children with poliomyelitis, cerebral palsy and other diseases that touched larger groups.
- In 1975 the centre was completely rebuilt and started slowly to arrange courses for more rare diseases like cystic fibrosis.
- From the mid-70s Frambu became the first institution to organise seminars for families with rare diseases.
- 1979, Oslo, 1st conference on rare diseases

Short history of Frambu (cont.):

- Several user associations and patient organizations were “born” or founded at Frambu during the 80s and later.
- The efforts and experiences from Frambu were recognized by the health authorities in Norway and contributed to the first national initiative on rare diseases in Europe, in 1980.
- The “rare field” became a health priority area from 1990-93.
- In 1996 Frambu was established as a Resource Centre for Rare Disorders, as the first and largest of the 16 (now 10) national resource centres for rare disorders in Norway.

Short status of Frambu:

- Frambu has a **national responsibility** within the rare field.
- Frambu is neither directly involved in investigations, diagnostic procedures, nor giving treatment, but indirectly we are often consulted. **Our service** is to give reliable and up-dated information and share evidence based experience.
- **Supplementary function** to the ordinary health care system through the specialised health services, offering **different types of services**, like arranging **courses** for families, siblings, grandparents, professionals etc., producing information material and giving information by having **videoconferences**, running health camps and performing **visits** to local users and families, communities, schools, sheltered homes, working places, etc.
- **Research and developmental work** are important growth areas and have become integrated parts of the activities of Frambu.

Short status of Frambu (cont.):

- Frambu is an **independant foundation**, but achieved to get its own fiscal position in the Norwegian State Budget, until 2007. Since then it has been part of the ordinary health care system, organised in the south-east health region.
- From 1st January 2014 Frambu is an **integrated part of the new umbrella organization for rare diseases and disorders in Norway (NKSD)**, as 1 of 10 centres, but still an independant foundation.
- Designated to be "responsible" for **more than 120 different**, rare disorders, diagnoses, syndromes and groups of disorders.
- **100% financed** by designated **governmental subsidies** from the Norwegian state.
- All services offered by Frambu are **free**. Stay and travel are free.
- The parents are getting **educational leave** (no sick leave) for receiving information and training during the courses.

Visions and goals for Frambu:

- To be a **meeting place** and a **learning place** for users, their families, care givers and support service system.
- To give reliable and **correct information** and **share experiences** about the diagnosis, its challenges and good solutions.
- In stead of contributing to resignation and despair: **Learn to live with!**
- **Empowerment:** Support and strengthen the users and their families in meeting and tackling the challenges of daily life in a better and more appropriate way!
- Contribute to a **more hollistic and equal follow-up** for persons with a Frambu-diagnosis all over the country, **through dialogueing with care givers and professionals in the interdisciplinary support service.**

Frambu Centre for Rare Disorders:

- 85 employees: 13-14 different professions: (doctors (pediatricians), psychologists, physiotherapists, occupational therapists, clinical nutritionists, special teachers and educational therapists, augmentative alternative communication (AAC) therapists, behaviour therapists, social educator, journalist, video journalist, photographer, communication and information technologists, social workers, etc.)
- Hotel facilities with 70 beds, grand kitchen, maintenance etc.
- Kindergarden and school with preschool teachers, teachers, milieu therapists, etc.
- **The Frambu model of “learning to live with”** has become a sustainable “export product” and is both a way of thinking and a solution that ought to be considered when developing and expanding services for people with rare diseases/disorders.

Courses for the Rett Syndrome community in 2015 to be arranged at Frambu :

- 1 RS specific week course for families (all ages)
- 3 week courses for families with overlapping focus within the "RS-field"/impairment, age-specific (0-3y, 5-16y, >18y)
- 3 courses (1-2d) for professionals working with people that have challenges/difficulties in communication and interaction; psychological aspects; transitional challenges from kindergarden to school
- 1 course (3d) for grandparents: "The good helpers"
- 1 course (3d) for next of kin/relatives of grown-up women with RS
- All courses may be joined and attended by video connection and conferencing all over the country.

- Frambu
- Rett syndrom

What will need attention?

- The child's well-being, growth and development.
- Symptoms and signs.
- Look for conditions described to be common or rare by the syndrome, like organic manifestations and so on.
- Be aware of preventable failure of development and be prepared to take measures so that it less likely comes true. (ex. scoliosis).
- Think broadly and simply! Syndromic children get everything that non-syndromic persons also may get!

Comorbidity

- = Conditions or diseases that may occur or appear together with the main diagnosis or the disorder, but may have another cause, explanation or treatment.
- Many of the rare disorders at Frambu have plenty of cormorbidities connected, that should be examined and investigated separately for what they add of extra burden of trouble and challenges, pain and concerns for the person affected. And several will even have options for treatment!

Other important areas and topics of attention and concern:

- Nutrition/eating problems, gastro-oesophageal reflux, etc)
- Sleep
- Motor function
- Speech and communication (augmentative alternative communication, AAC)
- Social development
- Emotional development
- Mental level and cognition
- Concentration and attention
- Activity (hyper-) and impulsivity
- Behaviour and aggression
- Challenging behaviour and selfmutilation
- Assessment of possible pain conditions
- Others? Etc.

Important elements in the follow-up of persons with one of the disorders of Frambu:

- A functioning "responsibility-group" around the child/person and her/his family
- An interested general doctor
- A stimulating kindergarden and school
- An including day care centre and/or working place
- Communication which is understandable and mutual
- Engaging and involving activities in leisure time
- Bodily exercise that takes care of physical and mental well-being
- Environment that protects and makes you feel safe
- Understanding caregivers and loving communities and accepting societies with equal legal rights to a good life



Thank you!

