7th European Rett Syndrome Conference

Day 1

# Collective Commitment to Rare Diseases

## Health data applied to rare diseases: review of the national plan for rare diseases 3 and prospects for the national plan for rare diseases 4 in 2024. Anne-Sophie LAPOINTE (France)

Context of rare diseases, obstacles: diagnostic error, limited information, geographic dispersion, access to treatment.

France had 3 rare disease plans so far. Third one being holistic, with care throughout the life span. 3rd plan is coming to the end now which had various ambitions: enable rapid diagnosis, innovate to treat, improving quality of life and autonomy of patients, structuring of date, (have a photo) etc… Guide, coordinate, share.

France also has care guidelines for rett syndrome (only mentioned it).

All speech is more orientated for rare diseases in general rather than RS.

Spain has two ERNs: ERN EpiCARE and ERN Transplant??

BNDMR – the French national rare disease registry data (photo) [www.bndmr.fr](http://www.bndmr.fr)

Plan 4: Focus on actions for neonatal screening, Innovations and treatments…

## PNDS or Rett center. Nadia BAHI-BUISSON (France)

National diagnostics protocol (PNDS). Was created bases on the extensive literature review and with a group of experts.

* Role of general practitioner. They need to be the heart of the treatment plan. They need to be able to redirect for specialists that rs patients need depending on their phenotype. Which ones are really needed and which ones are optional.
* Take into account puberty and change into adult life. It´s important to screen comorbidity and detect it early, to optimize their development, treat comorbidity and prevent situations of emergency.
* Graphic of the revisions (medical).
* Provides recommendations for certain problems, like motricity, respiratory disorders, etc.
* Breathing problems don´t have treatment (except sleep apnea). The aim of general practitionnaire is to detect these sleep apneas by poli and asses nutritional impact.
* Cardiac problems: 15% of rett girls have prolongation of QTc and reduced heart pace. Avoid medications (fluoxetine is mentioned directly). Or care with anesthesia.
* Gastro problems. Abdominal pain, bloating. GP needs to know all of this. It produces reflux, irritability, bad sleep, burps, etc.
* They suggest common treatment for each comorbidity in their protocol.
* Undernurishment. There are rett specific curves of growth (motil et al Pediatr Gastr Nutr 55:292 (2012). Gastrostomy if necessary. If the feeding takes longer than 30 minutes per meal then you should look into solutions, look for a eating, a dietary plan. Parents usually like that their girls chew, that they enjoy food, but you need to consider if she gets enough food because chewing is tireing. And why does she not swallow? Is she afraid, is there a physiological problem, are they chocking, are they coughing, etc. there´s a risk for lungs as well in the inhale food.
* Osteopenia. Its very common for complex disabilities and even more so if there is undernutrition. Increase vitamin d, calcium.
* Scoliosis. 80% have it over 35 years. Regular revisions. Many need surgery.
* Orthopedic problems. Hip dislocations are common, need to review systematically the joints. Train parents how to live with these problems because physio once a week is not enough, parents need to know how to relax and move joints. This could prevent some surgical interventions.
* Gynecological revisions. Periods are usually irregular, premature menstruations are common, regulation of period with medication is a sensitive topic because it can affect also the bones…?

Frequent commodities (list with percentages)

Rett Centers. They try to meet what parents see as most important for their child: communication, control of epilepsy, use of hands, walking, sleep, breathing, bowl movement. Thus, the objectives: developing enhanced treatment in multidisciplinary approach. It´s a “team sport”. It´s not enough to visit separate doctors, they need to gather together and coordinate with one being a lead. Its important to gather together and ask questions together to the patient, not separately.

Today there is no rett center really that exist in France. It’s a polihandicap center… ?