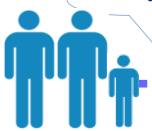
Patient Journey Rett Syndrome

Note: Most difficult and challenging time for the family, who believed their child to be healthy

Note: Symptoms and clinical presentation become more pronounced

Note: Intense therapeutic approaches could further improve the prognosis of Rett syndrome people



1 … symptoms and features of the disorder become apparent

2 … post diagnosis

3 … regular checks – x-rays and scans …

4 … referral to appropriate specialist when health need identified …

1. First Symptoms

Regression (9-30 months)
Some difficulty feeding
Floppy and poor coordination of limbs, trunk and stereotypic hand movements,
Walking, if achieved at all becomes unsteady,
Head circumference if normal at birth slows.
Onset of epileptic seizures
Abnormal breathing patterns emerge; breath holding, hyperventilation, air swallowing
Eating, chewing and swallowing problems
Heart rhythm abnormalities
Social withdrawal, confused (not autism).
Partial or complete loss of spoken language
Gastro intestinal problems

Note: Detailed care plan needs to be put in place to address the health needs of the specific comorbidities!
Rett Health Checklist available to download from www.rettuk.org ("Resources for families")

Ideally: Support for the family can be accessed from the experienced and professional national Rett organizations. Contact www.rettsyndrome.eu.
Access to knowledgeable and supportive GP.

2. Post Diagnosis

Need for involvement of multi-disciplinary team and co-ordination of care to manage symptoms with a care plan.
Referral to paediatric (child) or neurological consultant (adult)

Ideally: Care plan to involve physiotherapy, occupational therapy, SaLT, hydrotherapy and music therapy - can help a person with RTT lead a happy and full life.
Have an up to date Health Passport for use in hospital consultations or emergency

3. Surgery

Scoliosis
Removal of stones (gall bladder or kidney)
Other orthopaedic interventions, e.g. hip, feet
Gastro-intestinal surgery when symptoms become severe
Dental surgery often needed under anaesthetic

Ideally:
Regular checks including X ray or scans where appropriate to check on progression of any identified problems.

4. Follow-Up…

Problems may exist when passing from paediatric to adult health care services
Illnesses may be wrongly attributed to "having Rett syndrome," when in reality the condition is found in a neurotypical person of the same age and should therefore be treated in the same way.

Ideally: Annual health check with GP; include long QT check, blood tests, full body examination
Any symptoms identified should be promptly referred for further investigation and escalated up the care pathway where necessary
Use Rett Disorders Alliance UK Health Checklist to guide parents/carers and professionals through the complex symptoms and problems of the disorder.
Download from Rett UK website, www.rettuk.org

Information and European support groups:
www.rettuk.org - support helpline telephone +4401582 798911
www.rettsyndrome.eu

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Patient Journey Williams Syndrome

Note: Can experience poor growth, cardiac abnormalities!

1 … pre-diagnosis — 2 …. genetic analysis … — 3 — 4 — 5

Note: Often we don't know which our rights are, we don't know which chances we can seize because there is no information!

1. First symptom
Supravalvular aortic or pulmonary stenosis,
Low growth, characteristic Physiognomic aspect
Hypercalcemia
Feeding difficulties
Dental abnormalities
Hernias
Hyperacusis
Musculoskeletal problems
High urinary frequency - nocturnal enuresis

Ideally:
A centre of competence that could bring together all the symptoms asking to a geneticist to do specific analysis.
The capability to recognize symptoms and quickly give to parents a diagnosis.

2. Diagnosis & 1st Treatment
Heart controls (surgery if necessary)
Neuro - psychomotricity
Logopaedia
Physiotherapy
Music therapy

Ideally: The chance to do all the analysis in the same place;
Capability of communication to afford the first steps into the Syndrome;
Start the early intervention to accelerate the development (especially motion perception and micromanipulation.

3. Clinical & Mental Check
Clinical follow up (twice yr)
Table of growth specific for WS and guidelines about what to do at the different ages (cardiological controls, pressure, endocrinological controls, and many others)
Neurological FU (each yr) to evaluate the stage of mental development and the gap with the normal parameters

Note: Case Manager / Coordinator and Multi-Professional Approach!

Ideally:
All the therapy needed to express at the best all the potentiality of the person

4. Follow-Up
Program of mental improvement, attending to a neuro-psychotherapy x2 week until 6/8 years;
Speech therapy programme (from 2 to 4/5);
Music therapy all lifelong;
Educational program to improve their autonomy.

Note:
Coordination Centre to case manage a multi-professional team (teacher, therapists, doctors, family, educators….)
See MDT x twice yearly
Aim to improve capability and quality of life!

Ideally: Continuous development programme (e.g.: in a resource centre)

5. Social Care
Educational support at home to develop autonomy
Psychological support to afford anxiety and to implement awareness about "self" /who they are, which are their limits, why they can't do what other people are usually doing etc.).
Occupational therapy
Recreation programs

Ideally:
We need a well-structured social system that could give us a guideline about all the things needed. We should find answers to our questions and solution for our problems!

Information and European support groups:
www.eurowilliams.org

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Patient Journey Prader Willi Syndrome

Note: My little girl seemed unusually tired, respond poorly to stimulation, have a hard time waking up or have a weak cry!

... delayed motor development ...

1 — 2 — 3 — 4 — 5 — 6

... poor responsiveness ...

... genetic diagnosis

Note: Care for life ... happiness as part of the treatment!

... rehabilitation program everyday & low caloric diet ...

1. Pre-Diagnosis: Poor muscle tone; Distinct facial features
Poor sucking reflex and difficult feeding; Almost no cry.

1st Symptom: Hypotonia and difficulties in feeding;
Poor responsiveness; Sitting up and walking — later Hypotonia and difficulties in feeding; unusually tired, respond poorly to stimulation, have a hard time waking up or have a weak cry.
Sitting up and walking later.

Ideally:
Improvement of muscle tone
Special technics for feeding and stimulating exercises
Special technics for feeding

Medical treatment and early rehabilitation intervention

2. Diagnosis
Unusual food-seeking behaviors, hypogonadism
Intelectual disability;
Delayed motor development
Speech problems
Small hands and feet

Thick saliva, dental problems caused by gastric acid reflux.
Overweight if not eating low calorie diet

Note: Medication & rehabilitation
Support for education & school integration & inclusion into community

Ideally: GH Treatment
Sex hormone treatment
Permanent rehabilitation program; Education and work
Independent life skills

3. First Treatment
Delayed in motor development
Rehabilitation program, 3 weeks every 3 months

Surgery – some cases of scoliosis

Physiotherapy
Dietician

Ideally:

Improvement in general health and reduced weight

Information and European support groups:
www.apwromania.ro

4. Follow-Up Care...
Delayed in motor and intellectual development, behavior problems,
Speech problems
A special behavior. Easily stressed. Often anxiety with changes. Autistic symptoms , social interaction difficult some develop psychiatric diseases

Overweight and sleep apnea;
Medication & rehabilitation
Support for education & school integration & inclusion into community;
a proper job and qualification

Ideally:
Maintaining the weight, even reducing it
Relevant school education, individual approach.
NO one can ever live alone, need supervision of food and eating. Need much personal support to avoid behavioral problems.
Education and work. Cannot live independent, but many learn many skills
Work: must be sheltered and reduced hours: no one can work normally 8 hours per day. They do not have the strength.

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Patient Journey Spina Bifida

Note: Might be detected in the womb at an ultrasound control, defect on the spine or an unnatural big head.

Note: Patient will be at the hospital/clinic/specialist center for 4-7 days so it is possible to do all the exams!

1 … at 16-19 wks of pregnancy …

2 … at birth …

3 … diagnosis straight after birth …

4

5 … from birth to departure from life. First time when the baby is born. After that needs a checkup every year …

1. & 2. Pre-Diagnosis & 1st Symptom
The child is born with Spina Bifida which is showed as a malformation on the back. Information about Spina Bifida from HCP and a visit from another parent with a child with SBH after approx 4 weeks after birth. Depending on how the parents are coping.

***Note:** The parents can choose about proceeding with the pregnancy, surgery in the womb or leave it as it is and wait until birth!*

Ideally:
The parents feel their being well taken care of, having faith in the doctors. Being well informed on why and what to do next

2. Diagnosis
IF the malformation is low down on the spine = less damage

If the malformation is high up on the spine = more damage
Measure the head to look for hydrocefalus

Ideally:
The parents have a patient responsible doctor who has all the information on the child and who is listening to parents concerns

3. First Treatment
Surgery straight after birth within 24 hours.
Close up the back
If the patient has hydrocefalus.
Put in a shunt in the ventricle.

5. Follow-Up Care

Peadiatric Nurologist as Clinical Lead and Urologist and/or Urotherapeut,
Orthopedic,
Neurosurgeon and Radiologist

Note:
Urologist/urosergeon – checks up the kidney, bladder and bowl movement. If there are any problems a discussion is needed for surgery.
Urotherapist – examine bladder control and bowl movement. Teach how to do clean intermittent catheterization with is very important to be able to do yourself.
Orthopedic – checks for defects on feet's, knees, hips, scoliosis, kufosis, etc. If needed – consult with an orthopedic technician or surgery.
Neurosurgeon – Hydrocefalus, Arnold chiari syndrom, tethered cord which can do a lot of damage like lost of feel, pain and so on. If problems there is a need of surgery
The Neurologist – is the patient responsible doctor who checks out the rest like epilepsy, eyes pressure, cognition, ability to swallow and eat, weight (to big /to small) etc and writes referrals to specialists in the area for further treatment.

Information and European support groups:
https://www.ifglobal.org/

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Patient Journey Pitt-Hopkins Syndrome

Note: Information to parents and contact with support groups if desired!

… diagnosis can take many years …

Note: ideally we will find a treatment one day for the breathing anomalies and other ANS dysfunction

1 … usually normal pregnancy …

2

3

Note: Diagnosis explained many parents are given a print out of a medical paper and sent on their way!

4

5 … continuing need for physiotherapy and other therapies. …

1. & 2. Pre-Diagnosis: Usually normal pregnancy. Some ultrasounds may show something, some may have initial feeding problems but others not. Parents will begin to onotice delay in developmental milestones, as child grows.

1st Symptom: Lack of smiling at 6 weeks and all motor milestones usually delayed. Distinct facial features which although dysmorphic not strikingly. Hypotonia.

3. Diagnosis & 1st Treatment
Blood test only available since 2007 but needs a geneticist to suspect PTHS MRI may find some structural anomalies. Distinct facial features, motor and cognitive delay, hypotonia, lack of speech, breathing regulation anomalies, unstable gait if walking, GI problemsconstipation, reflux myopia, slender fingers, palmar crease, stereotypies

4. Surgery
Some children may need foot surgery if their feet pronate too much. Some may need surgery for undescended testes. Possibly for strabismus

5. Follow-Up
Many children need medication for constipation, reflux, seizures. Some have oxygen for breathing anomalies.

Ideally:
Doctors take parents' concerns seriously early so therapy can begin promptly. Initially this is physiotherapy.

Note: Physiotherapy once doctors agree there is developmental delay. In addition children often receive OT, ST, MT and SIT. Regular follow-up by paediatrician. May have medication for constipation or reflux

Note: Sight and hearing checked. Feet looked at and reviewed regularly for specalist footwear. Back checked for scoliosis. EEG done for base-line. Advice for constipation. OT assessment for equipment

Note:
Hydrotherapy available after surgery and other physiotherapy***!***

Ideally:
Good communication between different specialists and therapists.

Note: SEN schools involvement early on. Respite for family. Help with care in home. Regular access to specialist***!***

Information and European support groups:
www.pitthopkins.org.uk
www.pitthopkins.nl
www.pitthopkins.ch
www.aisph.it

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