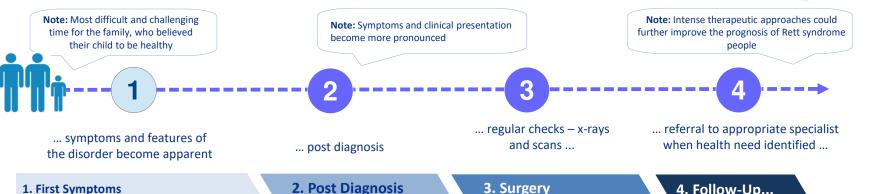


Patient Journey Rett Syndrome





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 Λ

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

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

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.

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.rettsvndrome.eu. Access to knowledgeable and supportive GP.

EURORDIS.ORG

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

Ideally:

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



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,

Ideally: Annual health check with GP;

include long QT check, blood tests, full

www.rettuk.org