Syndrome Specific Medical health check guide - Rett-Syndrome (RTT)

Introduction
Classic Rett-syndrome is an X-linked dominant disorder caused by spontaneous ‘de novo’ mutations in the MECP2 gene located on the X chromosome. It affects 1 in 12,000 girls born each year and is only rarely seen in boys. This gene provides instructions for making a protein (MeCP2) that is critical for neuronal function, whilst mutations in the MECP2 gene reduces the activity and communication of certain neurons. Pre-clinical research in mice strongly suggests that RTT could be one of the first curable neurological disorders.¹

Classic RTT is defined by a regression phase between the ages one and four years with partial or complete loss of spoken language, dyspraxic gait and stereotypic hand movements such as ‘hand mouthing’.

Although most women with Rett-syndrome reach adulthood, some will die at a fairly young age as a result of complications, such as heart rhythm abnormalities, pneumonia and epilepsy. There are currently only a few women surviving over 40 years.

History
As with all people with LD focus on an assessment of:

- eyesight and hearing
- feeding, bowel and bladder function
- behavioural problems.

Communication: Adolescents and adults are likely to rely on non-verbal communication. Periodic unexplained agitation, laughing or crying is common and may be associated with poor central parasympathetic restraint in RTT. A quiet and relaxed atmosphere helps it. Agitation is also the means to express any pain, irritation, discomfort, distress, anger, frustration or boredom and such causes must be carefully excluded. Sedatives and antipsychotics should be avoided. Short-term use of a serotonin reuptake inhibitor may be helpful in extreme cases.

Sleep disorder: May include failure to go to sleep, night-time waking and daytime sleeping. Active days help to ensure quiet nights, and bedtime routines are helpful. The individual should sleep alone with a ‘baby alarm’ if necessary and intervention should be

minimal. The room should be warm and safe to move about in without risk of injury. Melatonin may help to establish a routine.

Breathing rhythm is usually normal asleep and abnormal on alerting. Apneustic breathing (prolonged inspiration) occurs mainly in younger and Valsalva breathing in older women. Shallow breathing, breath holding and central apnoeas may lead to severe hypoxia.

Non-epileptic vacant spells are more frequent than epileptic seizures in Rett and are due to reduced brain stem cardio-respiratory control. This may lead to episodes of loss of consciousness, which may be difficult to differentiate from epilepsy and may require concurrent monitoring of central autonomic function with electroencephalography. Vagal tone and baroreflex sensitivity are usually reduced.

Dystonic spasms are common. Gentle massage may be more effective than medication. Osteoporosis has been reported in Rett, even in active people.

A balance must be found between providing active movement, which is essential for health and adequate protection from trauma.

**Examination**

1. **Sensory**
   - Full assessment by optician/optometrist every 2 years
   - Assessment and refer for Audiology assessment if concerns

2. **Dental**
   - Dental Check for grinding (bruxism) and ensure regular tooth cleaning and visits to the dentist.

3. **Gastrointestinal**
   - Poor feeding may be due to postural problems and reflux is common. There may be aerophagy which commonly accompanies the abnormal breathing. Very severe cases may be helped by per-cutaneous gastrostomy.
   - Examine the abdomen for constipation and abdominal distension

4. **Neurological**
   - Central Nervous Epilepsy is present in about 50% and may remit. Generalised or partial seizures respond to medication according to type. Since the EEG may be epileptogenic
without clinical epilepsy, video during prolonged recording may be necessary to distinguish epilepsy from non-epileptic vacant spells (see above). Hand stereotypy is involuntary and increased by alerting. It can be ignored unless injury occurs, when a light elbow splint may be used to prevent injury with minimal interference. Task performance may improve with one hand gently held (only during the task).

☐ Check seizure control and medication at each visit. Expect to wean off anticonvulsants if seizures become infrequent.

5. Cardiovascular
☐ Examine the feet and legs for poor blood circulation to the lower legs and feet (vasomotor disturbances). Consider sympathectomy if severe.

6. Musculoskeletal
Posture and joint position are likely to deteriorate due to initial hypotonia and later hypertonia.

☐ Review posture and joint position. Large joints of shoulders, hips, knees and ankles are at risk of permanent flexion or extension in fixed postures (joint contractures).

☐ Assess for Scoliosis, which is common and deteriorates during growth spurts. Ensure the person is receiving postural care and refer to orthopaedic surgeons for more severe or progressing curves.

☐ Assess hand skills, which are usually poor (dyspraxic) but improve given opportunity and encouragement. Gentle massage of the hands just before a task may encourage use e.g. holding mug or spoon within the adult’s hand in feeding.

Resources
Rett UK resources, including Management Guidelines: http://www.rettuk.org/resources/

Useful particularly for describing the 4 stages of Rett http://www.nhs.uk/conditions/rett-syndrome/Pages/Introduction.aspx