Rett Disorders Alliance UK

RETT UK • REVERSE RETT • FOXG1 UK

Rett Syndrome Health Checklist

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Introduction

What is Rett Syndrome?

Rett Syndrome is a rare neurological disorder affecting mainly females and very few males. It is present from conception and usually remains undetected until developmental delay occurs around one year of age, when children may lose acquired skills and become withdrawn. It should be noted though that some children may regress later and/or have minimal regression. Rett Syndrome is usually caused by a fault (mutation) on a gene called *MECP2* which is found on the X chromosome. In 99% of cases it is a new mutation and not inherited. There are many different types of mutation and this can impact on clinical severity seen in patients. People with Rett Syndrome usually have profound and multiple physical and communication disabilities, and are totally reliant on others for support throughout their lives

What is the Rett Syndrome Health Checklist?

The Rett Syndrome Health Checklist provides families, carers and health care professionals with an overview of i) the multiple simultaneous health problems that typically occur, (ii) the warning signs that would alert you to the different problems and (iii) referral and treatment pathways, plus links for further reading including relevant research papers.

How was the Rett Syndrome health checklist developed?

The checklist has been written by the Rett Disorders Working Group. This group includes experienced Rett specialists in primary and secondary health care as well as parents and representatives from the wider patient organisations (Reverse Rett, Rett UK and FOXG1). Please see list of acknowledgements regarding the professionals who have been involved and their disciplines.

Key Principles for Primary and Secondary Health Care Professionals

The Health Checklist is not intended to be an exhaustive list. Whilst we know so much more about Rett Syndrome than we did 30 years ago, we are learning new things all the time, particularly about how the syndrome manifests in middle age, as we now have the first people who were diagnosed in the UK reaching middle age and later life.

Having a diagnosis of Rett Syndrome does not exclude the person from developing other problems that you would see in the neurotypical population, but symptoms may be masked by the complexity of the disability and the person's limited ability to communicate. Talking and listening to parents and carers who know the person really well is key to understanding the nature of the problem and what is 'typical/atypical' behaviour or presentation for that individual.

Starting point for GPs

- Is this symptom normal in a neurotypical person of the same age?
- Is something new, or has something got worse?
- Has this led to increased impairment?
- Could any medications/contra-indications be responsible?

Be honest about the limitations of your knowledge and recognise the knowledge of the parent or carer – both about the condition and their daughter/son/client. Escalate up the care pathway where necessary to ensure accurate diagnosis and treatment.

If new medications are to be introduced, this should be started from a low dose and continued at the lowest dose at which beneficial effects are seen. Any increases necessary should be implemented slowly with careful monitoring of all symptoms for any adverse effects.

Stages of Rett Syndrome

Rett Syndrome has been described as being in four stages: early signs, regression, plateau and late motor deterioration (Refer to NHS Choices Rett Syndrome). This can be applied in general terms but there is a spectrum in severity and environmental factors can influence the progression of the disorder to some extent. Some symptoms fluctuate as the disorder progresses. Some children who are at the severe end of the spectrum may have a less marked regression as they did not develop many skills in infancy, because problems emerged earlier on. Some continue to ambulate throughout life.

However, what should be noted about the fourth stage instead, is that it does not imply an end of life scenario and the same investigations, treatment and care should be instigated as they would be for a neurotypical person presenting at A & E or as an inpatient.

Rett 'episodes'

Some Rett literature, particularly the less recent information, refers to ‘Rett episodes’ as a general way of describing an unusual movement or breathing pattern but this is not helpful in finding the root cause of the problem and treating it appropriately. We strongly discourage the use of this term. Every effort should be made to accurately assess and diagnose the problem.

Annual Health Checks

Young people with Rett Syndrome aged 14 and above should have at least an annual health review with their GP. Whilst their care is likely to be managed by a community paediatrician up to age 18, it is very important that the GP does have some knowledge and experience of the young person as they will become the first point of referral post 18.

‘Survival in to the 5th decade is typical in RTT, and death due to extreme frailty has become rare.’ (Tarquinio et al)

In a recent study entitled, ‘*The changing face of survival in Rett Syndrome and MECP2-related disorders,*’ survival for classic and atypical RTT was greater than 70% at 45 years.

Whilst Rett Syndrome can be life-limiting, the leading cause of death is cardiorespiratory compromise. Many of these risk factors for early death can be managed. Intense therapeutic approaches could further improve the prognosis for patients with Rett Syndrome.

Cognitive Ability of People with Rett Syndrome

Parents and carers for many years have said that their daughter/son understands far more than they are given credit for but until very recently there had been few attempts to try and assess this. Advances in eye gaze technology, in particular, have facilitated some meaningful assessments of language and cognition in children with Rett Syndrome.

For example, in studies published by Clarkson et al. (2017) and Ahonniska-Assa et al. (2018), some children were found to have a significant learning disability, others were found to have levels of understanding appropriate for their age, and one or two were above average for their age.

As with the range of symptoms in Rett Syndrome, we are now seeing there is also a range of cognitive ability which may also be influenced by their environment to some extent, but the important message here is not to assume they do not understand. Find out how they communicate, give them every opportunity to be included in conversations, give them the opportunity to express their opinions and wishes. In other words, presume potential.

References:
Clarkson T, LeBlanc J, DeGregorio G, Vogel-Farley V, Barnes K, Kaufmann WE, et al. Adapting the Mullen Scales of Early Learning for a standardized measure of development in children with Rett Syndrome. Intellectual and Developmental Disabilities. 2017;55(6):419-31. Ahonniska-Assa J, Polack O, Saraf E, Wine J, Silberg T, Nissenkorn A, et al. Assessing cognitive functioning in females with Rett Syndrome by eye-tracking methodology. European Journal Of Paediatric Neurology. 2018;22(1):39-45.

Key Principles For Parents

You know your son/daughter best and you are their best advocate. Often families of people with Rett Syndrome have significant knowledge and understanding of the disorder. This checklist will support you in conveying critical and validated information quickly to health professionals who may not have experienced Rett Syndrome before.

Keeping a diary and taking video recordings are a very helpful way of tracking changes in behaviour and/or symptoms that you can share with health care professionals to help them to understand your concerns and help with a diagnosis and treatment. Video recordings of when the person is well, as well as of symptoms, can also be useful.

Having an updated record of all medications, current treatment plans and past interventions/surgeries etc. is very helpful to share with health care professionals and saves you having to remember/repeat all this information during consultations or emergency admissions. A good way of doing this is to use either the Rett UK Health Passport or the Reverse Rett Hospital Passport.

The Health/Hospital Passports will be updated regularly so please refer to Rett UK or Reverse Rett websites to check for updated information. Many families find it really helpful to connect with other families through social media (there are two groups in the UK; Rett Syndrome Facebook Group for UK Families and Rett Syndrome Group for UK Families and Professionals). Both Rett UK and Reverse Rett offer a variety of events where families can meet in person.

Advanced care planning

Having an advanced care plan may also be useful to think about what you would want to happen in certain circumstances and have this clearly documented.

Advance care planning is normally a process of discussion between the patient and those who provide care for them, for example, the nurses, doctors, care home manager or family members. Depending on the capacity of the individual with Rett Syndrome, they may choose to express some views, preferences and wishes about their future care. Where this is possible, every reasonable effort should be made to allow them to be involved in decisions which affect their future. However, we recognise that for the majority of people with Rett Syndrome it will be their family and people who know them very well who will be making these decisions.

NHS Choices have a good booklet about Advance Care Planning and Rett UK have some useful guidance on their website.

Rett UK is a national charity dedicated to supporting and empowering people with Rett Syndrome and their families.

Our strategic aims are to…

- Provide professional family-support services, activities and events at a local, regional and national level.
- Educate health, social care and education professionals about Rett Syndrome to enable quicker diagnosis and improved standards of care throughout the UK.
- Advocate for people with Rett Syndrome to be given the support and resources needed to be active communicators.
- Promote, support and encourage research into new therapies and treatments for Rett Syndrome.

Our vision…

That everyone with Rett Syndrome is given every possible opportunity to achieve their individual potential so that they may live their life to the full.

www.rettuk.org.uk

Reverse Rett is a UK medical research charity working to accelerate treatments and a cure for Rett Syndrome and related *MECP2* Disorders.

We do this primarily by:

- Funding research focused on treatments and cures.
- Facilitating the delivery of UK clinical trials and access to emerging treatments.
- Collating information from experts in the field and providing access to it.
- Running the UK's only **Rett Syndrome Patient Registry**

Our vision...

Our vision is a world where children with Rett Syndrome are diagnosed much quicker and earlier than they are at the moment. A world where we know much more about the progression of the condition and the factors that affect that progression. A world where at every stage of development, treatments are available to mitigate against the distressing symptoms which can occur and, ultimately, a world where treatments are available which cure the condition and allow people with Rett Syndrome to live normal lives.

FOXG1 UK

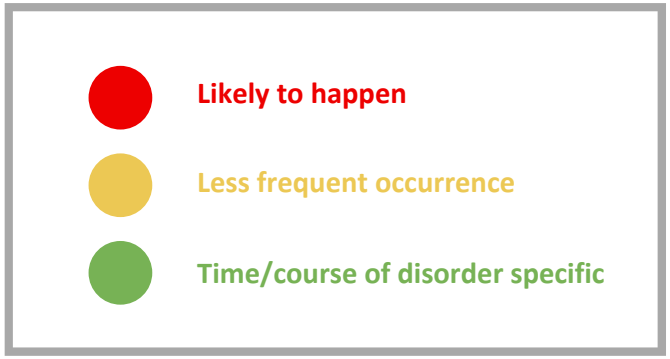
FOXG1 Syndrome has previously being referred to as Congenital Variant Rett Syndrome. It is now considered to be a separate condition with symptoms that are similar to Rett Syndrome but that are present from birth. Unlike Rett Syndrome it is found in both boys and girls. It is a very rare condition. Children diagnosed with the syndrome have a wide spectrum of abilities.

As FOXG1 Syndrome is a relatively newly identified genetic condition, there are no long term studies into the health or longevity of people diagnosed with the syndrome. For a few children it is clearly a life-limiting condition but there is every reason to believe that the majority will go on to live life to the full.

Clinicians who have knowledge of FOXG1 Syndrome are as rare as the syndrome itself, so families affected by this condition will also benefit from having access to the Rett Disorders Health Checklist.

FOXG1 UK are grateful to be part of The Rett Disorders Working Group.

Checklist



Respiratory

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Autonomic Dysregulation - Breathing Difficulties	*Central:* Apnoea/ breath holding, air gulping leading to abdominal distension (may lead to pseudo-obstruction). Hyperventilation and breathing holding typical of 'forceful breathers.' Feeble Breathing. Valsalva Manoeuvre. *Peripheral:* Bronchoconstriction is **NOT** part of Rett Syndrome – think Asthma! Use bronchodilators as per usual asthma treatment guidance.	Identify breathing type through full autonomic assessment with sleep study, video telemetry and EEG. Request advice to anaesthetists with specific reference to breathing irregularity identified. (People with Rett Syndrome of all ages often undergo emergency/planned surgeries). Hypotonia arising from treatment for other complications of Rett Syndrome may lead to reduced intercostal muscle tone. Full autonomic assessment to ascertain breathing type and treat accordingly. Buspirone may be helpful for some forceful breathers. Acetazolamide has been used in children with apnoeas. Feeble breathers often need oxygen support overnight. For autonomic assessment refer to: Dr Adrian Kendrick, Consultant Clinical Scientist, Department of Respiratory Medicine, University Hospitals, Bristol.	MacKay et al. Autonomic breathing abnormalities in Rett Syndrome: caregiver perspectives in an international database study. J Neurodev. Disord. 2017 Apr 28: 9:15 Singh and Santosh. Key issues in Rett Syndrome: emotional, behavioural and autonomic dysregulation (EBAD) - a target for clinical trials. Orphanet Journal of Rare Diseases. 2018 Jul 31;13(1):128. MacKay et al. Respiratory morbidity in Rett Syndrome: an observational study. Dev Med Child Neurol. 2018 Sep;60(9):951-957 Smeets et al. Management of a severe forceful breather with Rett Syndrome using carbogen. Brain Dev 2006; 28: 625-632 Julu et al. Characterisation of breathing and associated central autonomic dysfunction in the Rett disorder. Arch Dis Child. 2001 Jul;85(1):29-37 Andaku et al Buspirone in Rett Syndrome respiratory dysfunction Brain and Development, Volume 27, Issue 6, 437 - 438

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Aspiration *(The accidental inhalation of food, fluid or reflux into the lungs)*	- Arching or stiffening of the body during feeding - Irritability or lack of alertness during feeding - Refusing food or liquid - Failure to accept different textures of food - Long feeding times - Difficulty chewing - Difficulty swallowing - Difficulty breast feeding in infants - Coughing or gagging during meals - Excessive drooling or food/liquid coming out of mouth or nose - Difficulty co-ordinating breathing with eating and drinking - Increased stuffiness during meals - Gurgly, hoarse or breathy voice quality - Frequent spitting up or vomiting - Recurring pneumonia or respiratory infections - Less than normal weight gain or growth	Lower respiratory tract infection is the most common cause of death in Rett Syndrome. Prevention of chest infections is key to longevity in patients with Rett. Comprehensive multi-disciplinary feeding assessment to ensure correct posture for eating and drinking will help avoid aspiration. Video-fluroscopy/barium swallow if aspiration or unsafe swallow is suspected. Check for reflux symptoms and address actively. **Inform carers of precautionary steps:** Maintain 45 degree positioning during meals/eating/drinking/tube feeding and for 30 mins after. Minimal distraction during meal times. Enable patient to control speed of meal. Good oral hygiene, including teeth brushing and clearing mouth including pockets of mouth and leftover food around the mouth and lips.	Tarquinio et al. The Changing Face of Survival in Rett Syndrome and MECP2-Related Disorders. Pediatr Neurol. 2015;53(5):402–411. doi:10.1016/j.pediatrneurol.2015.06.003 Reverse Rett Aspiration in Rett Syndrome Fact sheet

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Silent aspiration	While overt aspiration can cause sudden noticeable symptoms, in silent aspiration, t**here is no coughing or clearing.** - Red watery eyes - Colour changes to skin around the eyes - Drooling - Changes in breathing/voice - Splayed hands in younger children		
Chest infections /Pneumonia	Prevention of chest infections is key to the longevity of patients with Rett. Early management of active infections leads to better outcomes for this patient population.	Annual flu vaccination for patient and carers. PPV/Pneumovax for patients over the age of 2 because of long-term respiratory issues and poor immune system. A second dose may not be neccessary. Input from respiratory consultant. Regular checks by specialist chest physio.	Kida et al. Pathogenesis of Lethal Aspiration Pneumonia in Mecp2-null Mouse Model for Rett Syndrome. Sci Rep. 2017;7(1):12032. Published 2017 Sep 20. doi:10.1038/s41598-017-12293-8
Preventative strategies	Regular chest infections, swallowing difficulties, aspiration. Increased secretions. If coughing becomes unusually frequent, observe carefully and take temperature regularly.	Use of prophylactic antibiotic during winter months may be useful. Daily mucodyne (carbocysteine) to keep secretions more fluid and easier to cough up. If temperature is raised, the patient appears unwell and or secretions are difficult to clear, notify GP and chest physio immediately.	

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Managing active infections	Raised temperature. Increased secretions. Generally unwell. Emergency anti-biotics specifically for active chest infections can be stored at home for immediate use in line with the individual patient's specified respiratory protocol, in order to avoid inevitable delays.	If chest infection is confirmed, stop prophylactic anti-biotics if in place and start **rescue anti-biotics** immediately. Increase dose of Mucodyne as appropriate. Use of nebuliser for saline or salbutamol inhalation as needed. Ensure regular position changes with alternate side lying particularly during the night. Sitting upright whenever possible to assist breathing. Chest physio by physiotherapist and trained staff. Monitor closely for any signs of deterioration, taking temperature regularly until condition improves. If there is marked deterioration, do not hesitate to take emergency action (111/999 call).	Don't leave it to chance article -Yvonne Milne MBE

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Sleep disturbance	Initial insomnia, nightmares, night terrors, nocturnal screaming/laughing. Obstructive central sleep apnea.	Basic sleep hygiene measures should be implemented. Parents/carers to record videos of breathing and sleep patterns to effectively highlight issues to sleep teams. Melatonin can be trialled in a general setting. A low dose of Clonidine can be used as a second line. Refer for full polysomnography to identify the route of the night time awakening. CPCP for Obstructive Sleep Apeoa NIV for OSA/CSA as appropriate and based on outcome of polysomnography. These treatments should be tried before or in conjunction with drug therapy. Avoid benzodiazepines, promethazine, chloral hydrate – may risk autonomic instability. Chloral hydrate use is to be discouraged and should only be used for the primary purpose of sleep induction under exceptional circumstances on a short-term basis, and under direct expert guidance. In children aged 2-11 years treatment with Chloral hydrate should be as an adjunct to behavioural therapy and sleep hygiene management, and usually for duration of less than 2 weeks. Long term use of chloral hydrate carries a high risk of serious digestive adverse events, sleep apnoeas, hepatic damage, respiratory arrest, cardiac arrythmias, dependency and withdrawal syndrome, and cancer. Use with caution and only under medical supervision. Refer to Cerebra Sleep Service, a free service for families.	Wong et al. The trajectories of sleep disturbances in Rett Syndrome. J Sleep Res. 2015 Apr;24(2):223-33. Young et al. Sleep problems in Rett Syndrome. Brain Dev. 2007 Nov; 29(10): 609–616. Cerebra Sleep Information Resources

Cardiovascular

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Long QT	An association between Rett Syndrome and Long QT has been noted, which may arise more commonly with increasing age. There are multiple meds which can increase QT interval. Common drugs to avoid: Fluoxetine, Azithromycin. Entromycin with other meds can be lethal.	Baseline ECG should be undertaken at diagnosis. ECG should be undertaken where there is suspicion of ECG abnormalities from clinical history and reported observational history. Repeated ECG where there are changes in the health status of the individual patient. ECG should be undertaken where there is desire to prescribe medication which is known to potentially cause QT changes with 48 hour ECG recording obtained prior to medication change. Where negative, once treatment is prescribed, repeat ECG should be undertaken for seven consecutive days. (May be adjusted depending on rapidity of action of the drug). If positive for ECG changes and/or prolonged QT interval, drug should be withdrawn and ECG recording continued to ensure return to pre-treatment status. Where possible, avoid drugs which prolong QT interval, where there is alternative available. ECG should be conducted as part of pre-surgical assessment especially when anaesthetic is being given. Website list/smartphone app for drugs to avoid with Long QT: https://crediblemeds.org/.	Ellaway et al. Prolonged QT interval in patients with Rett Syndrome. Arch Dis Child. 1999 May; 80(5): 470–472

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Cardiovascular dysregulation	Dysregulation of cardiovascular parameters. Heart rate, BP, peripheral vasoconstriction, may change markedly within seconds.	Monitor for QTc prolongation at least annually. QTc may vary markedly within a short space of time. Beware of/ avoid medications causing QTc prolongation.	Julu & Engerstromm, Assessment of the maturity-related brainstem functions reveals the heterogeneous phenotypes and facilitates clinical management of Rett Syndrome. Brain Dev. 2005 Nov;27 Suppl 1:S43-S53. Epub 2005 Sep 2 Kumar et al. Cardiovascular autonomic dysfunction in children and adolescents with Rett Syndrome. Pediatr Neurol. 2017 May;70:61-66.
Circulation	Very poor flow to extremities especially lower leg/feet. Risk of severe chilblains.	Doppler Scan. Thermal socks/footwear/foot spa. Warming slowly when cold, elevating feet. Consider Amlodipine for severe cases before considering GTN patches to decrease risk of severe headaches. Severe cases consider sympathectomy. Glyceryl trinitrate (GTN) patches may be helpful.	Kumar et al. Cardiovascular autonomic dysfunction in children and adolescents with Rett Syndrome. Pediatr Neurol. 2017 May;70:61-66.

Gastroenteric

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Growth and nutrition	Poor weight gain, prolonged feeding times, poor fluid intake, chewing and swallowing difficulties. Needs closer monitoring during adolescence when feeding problems can worsen.	Height, weight and BMI should be monitored regularly. Growth charts for North American populatons of people with Rett Syndrome are available here: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3468773/ Consider NG tube or Gastrostomy (with fundoplication if severe reflux) to supplement oral feeding if needed for weight gain and for adequate fluid intake. It is important not to feed a person with Rett with the aim of getting weight up to population mean for her or his age. Weight should correspond to mean weight for height/Rett specific growth charts. (link under further reading) Overfeeding can lead to obesity or challenges with gut being challenged by too much food intake leading to gut functional problems. Be vigilant about coughing/choking on food and drink. Think aspiration. Page 2.	Improving health outcomes in Rett Syndrome: Nutritional and Digestive Health. An information booklet for families and carers Motil et al. Gastrostomy Placement Improves Height and Weight Gain in Girls with Rett Syndrome, Journal of Pediatric Gastoenterology and Nutrition 2009: 49)2): 237-242
Gastroenterology	Difficulties include *reflux, diarrhoea (SNS overactivity), constipation/ pseudo obstruction (PNS overactivity) associated pain, vomiting, dysmotility. Watch for aspiration pneumonia in severe reflux (may lead to ARDS). Failure to empty bowels completely especially in older people. Impacted bowels – diarrhoea may be sign.	Movicol, Lactulose, suppositories may help with constipation as well as natural remedies such as prune juice. Use ARDS guidelines should this occur.	Helen Leonard Gastro-intestinal disorders in Rett Syndrome Checklist for clinicians on assessment and management Motil et al. Gastrostomy Placement Improves Height and Weight Gain in Girls with Rett Syndrome, Journal of Pediatric Gastoenterology and Nutrition 2009: 49)2): 237-242 Strati et al. Altered gut microbiota in Rett Syndrome, Microbiome. 2016; 4: 41

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Eating and drinking difficulties *Nervous System*	Dysphagia: Feeding, chewing, swallowing difficulties – length of time to eat, coughing, gagging.	Feeding and behavioural strategies; small, frequent and or thickened feeds. Eliminating selected foods from diet. Correct posture is critical for safety and for success with adequate oral nutritional/fluid intake. Good posture could include 'chin tuck' achieving a better position of the head to protect the airway. Combined assessment with multi-disciplinary team should take place in order to establish safe posture for effective eating and drinking with on-going support and review. Upright position while eating/elevating bed head to avoid aspiration. Video fluoroscopy, Barium Swallow Consider NG tube or Gastrostomy (with fundoplication if severe reflux)	

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Reflux Reflux occurs in 40% of children and adults with Rett Syndrome and must be managed due to the increased risk of aspiration. Reflux is also more likely if the person has scoliosis (common in Rett) or spends a lot of time in supine position.	Pneumonia/RTIs are the leading cause of death in Rett. Families and caregivers should be informed about the dangers of reflux aspiration and the importance of treating this and postural management. **Looking for:** Regurgitation Sour smelling burps or vomiting Dental erosion Unexplained weight loss Iron deficiency anaemia food refusal and/or ruminatio Recurrent lower respiratory tract infections Behaviour problems including agitation Self-harm Screaming Restlessness for no apparent reason	**Options to test for reflux include:** 24 hr oesophageal pH monitoring (with preference for conducting a multi-channel intraluminal impedance study in combination with pH testing). Upper GI endoscopy to assess whether there is reflux oesophagitis and/or gastritis. Radionucleotide scintigraphy to test for aspiration due to reflux. **Management:** Feeding and behavioural strategies; small, frequent and or thickened feeds. Eliminating selected foods from diet. Upright position whilst eating and for minimum of 30 mins after. Elevate bed head to 45 degrees for patients who are tube fed/receiving fluids over night or in bed. **Pharmacological management:** Proton pump inhibitors (PPIs, e.g. Lansoprazole, Omeprazole, Pantoprazole) are recommended as the drugs of choice. Prokinetics should be used with extreme caution or avoided in people with Rett syndrome because of their effect in prolonging QT interval. 20% of patients have a prolonged QT as part of Rett Syndrome.	Website/Smartphone app to check drugs for QT prolongation here: https://crediblemeds.org/

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Gallbladder dysfunction/ Pancreatitis	Pain/discomfort but can be masked. Fever, jaundice and vomiting. Diarrhoea, weight loss. Upper abdominal pain that radiates into the back; it may be aggravated by eating, especially foods high in fat. Swollen and tender abdomen. Nausea and vomiting. Fever. Increased heart rate.	Check for cholecystitis, gallstones and gallbladder sludge. After excluding gastroesophageal reflux, gallbladder disease should be considered as a cause of abdominal pain in RTT and cholecystectomy recommended if symptomatic. Physical examination, MRI, CT scan and ultrasound.	Freilinger et al. Prevalence, clinical investigation and management of gallbladder disease in Rett Syndrome, Dev Med Child Neurol. 2014 Aug;56(8):756-62

Nervous System

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Early developmental impairment (NB falls into diagnostic criteria and seen in early development)	Early significant delay is not typical for classic Rett but does occur in the congenital variant. Typical Rett will have some relatively minor delays but have a clear period of regression where skills are lost.	Documentation of milestones - gross motor, fine motor, speech comprehension, socialisation. Referral to appropriate services. Check vision and hearing.	Neul et al. Rett Syndrome: Revised diagnostic criteria and nomenclature. Annals of Neurology 2010; 68: 944-950 Neul et al. Developmental delay in Rett Syndrome: Data from the natural history study. J Neurodev. Disord. 2014; 6 (1): 20 Marschik et al. Early development in Rett Syndrome- the benefits and difficulties of a birth cohort approach. Developmental Neurorehabilitation 2018; 21: 68-72
Muscle Tone and Tone Management	Variable from low to high tone but frequently outside the normal range. See also autonomic dysfunction – hypertonia.	Referral to physiotherapy. Holistically assess with MDT for neurological disorders. Check positioning – standing (frame), appropriate supported seating, lying (sleep system). Check for co-morbidities (constipation) and medications (Benzodiazepines) that alter tone positively or negatively. Massage and stretching of muscle groups should be part of a comprehensive postural care plan (for people with Rett of all ages)) under the guidance of a therapy team. Pharmacological tone management.	Lotan, M., Merrick, J. (2011). Rett Syndrome: Therapeutic interventions. Nova Science Publishers. Lotan, M. (2006). Rett Syndrome, Guidelines for Individual Intervention. The Scientific World Journal, 6(6), 1504-1516. doi:10.1100/tsw.2006.252

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Dystonia	Focal or generalised - involuntary spasms and contractions. The movements are often repetitive and take on unusual and awkward postures. They can also be painful. See also autonomic dysfunction - hypertonia	Typical medication used for dystonia: Trihexyphenidyl, baclofen, gabapentin, clonidine, benzodiazepines. NB – There is no strong evidence base to guide this: Typical medication used for managing spasticity: baclofen, gabapentin, benzodiazepines, tizanidine	Yuge K et al (2017). Ghrelin improves dystonia and tremor in patients with Rett Syndrome: A pilot study. J Neurol Sci. 2017 15;377:219-223. doi: 10.1016/j.jns.2017.04.022. Humphreys P, Barrowman N. The Incidence and Evolution of Parkinsonian Rigidity in Rett Syndrome: A Pilot Study. Can J Neurol Sci. 2016;43(4):567-73.
Spasticity		Caution with benzodiazepine use, given risks of increasing secretions, respiratory depression, tolerance and dependency. Botulinum toxin can be considered for focal elements of high tone (either spasticity or dystonia).	Kadyan V et al. Intrathecal baclofen for spasticity management in Rett Syndrome. Am J Phys Med Rehabil. 2003;82(7):560-2. Temudo T et al. Movement disorders in Rett Syndrome: an analysis of 60 patients with detected MECP2 mutation and correlation with mutation type. Ramos E, Dias K, Barbot C, Vieira JP, Moreira A, Calado E, Carrilho I, Oliveira G, Levy A, Fonseca M, Cabral A, Cabral P, Monteiro JP, Borges L, Gomes R, Santos M, Sequeiros J, Maciel P. Mov Disord. 2008 Jul 30;23(10):1384-90
Altered Tissue Properties	Over time, any abnormal muscle contraction can give rise to changed tissue properties – with changes in the viscoelasticity (viscosity – measure of resistance to stretch, elasticity – tendency to return to original shape/length) that make muscles feel stiffer. This ultimately leads to fixed musculoskeletal deformity.	Close therapy review regarding risk of development of fixed musculoskeletal deformity, with prompt referral to orthopaedic surgeons if concerns arise.	Terence D et al. Classification and Definition of Disorders Causing Hypertonia in Childhood. Sanger, Mauricio R. Delgado, Deborah Gaebler-Spira, Mark Hallett, Jonathan W. Mink Pediatrics Jan 2003, 111 (1) e89-e97; DOI: 10.1542/peds.111.1.e89

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Epilepsy	May be present in up to 70% of people with Rett Syndrome but majority of people with Rett will have abnormal EEG. Often starts around age 4/5 years. Huge variation in type/frequency of seizures including gelastic seizures.	First hand description +/- video recording to make a confident diagnosis of epilepsy but 48 hour EEG with video telemetry and sleep study at baseline, including neurophysiological, cardiological and respiratory measurements at minimum. Seizures triggered by hypoxia/sleep disturbances may be better managed by primarily addressing these triggers. AEDS based on type of seizure. Recommended good practise for children and young people with epilepsy to have a person specific epilepsy care plan. See guidance in further reading. Ketogenic diet can be helpful for some. All children and young people with Rett Syndrome and refractory seizures should be able to access assessment for potential epilepsy surgery. It is unlikely that resective surgery will be appropriate, but Vagal Nerve Stimulation may be of potential benefit.	Krajnc N. Management of epilepsy in patients with Rett Syndrome: Perspectives and considerations. Ther Clin Risk Manag. 2015; 11: 925–932. Glaze et al. Epilepsy and the natural history of Rett Syndrome Neurology. 2010;74(11):909–912. doi:10.1212/WNL.0b013e3181d6b852 Tarquinio et al. Longitudinal course of epilepsy in Rett Syndrome and related disorders Brain. 2017;140(2):306–318. doi:10.1093/brain/aww302 NICE guidance: Epilepsy in children and young people.

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Paroxysmal episodes not due to epilepsy	May be linked to autonomic dysfunction.	48 hr EEG with video telemetry and sleep study to distinguish from seizures.	Haywood, P. et al. P037 Dysautonomia presenting as non epileptic seizures in Rett Syndrome European Journal of Paediatric Neurology, Volume 13, S32 D. G. Glaze et al. Rett Syndrome: characterization of seizures versus non-seizures. R. J. Schultz, J. D. Frost Electroencephalogr Clin Neurophysiol. 1998 Jan; 106(1): 79–83.
Movement Disorder and Motor Planning	Disturbance in motor planning that impacts on all purposeful tasks and movements.	Referral to Occupational Therapy and Physiotherapy. Verbal Cueing, avoid direct questions and instructions, acknowledge time needed to respond. Difficulties with motor planning can impact the ability to change from one position to another. Comprehensive physio plan involving all caregivers and professionals re postural transitioning should be put in place in order to maintain independence.	Lotan, M., Merrick, J. (2011). Rett Syndrome: Therapeutic interventions. Nova Science Publishers. Kitty-Rose R. Foley et al. Gross motor abilities and interventions in girls and women with Rett Syndrome: a literature review; longitudinal video analysis of gross motor abilities of girls and women with Rett Syndrome. Edith Cowan University Downs J et al. Perspectives on hand function in girls and women with Rett Syndrome. Dev Neurorehabil. 2014;17(3):210-7.

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
EBAD – Emotional, Behavioural and Autonomic Dysregulation (Emotional – Assess changes from baseline) Anxiety/panic	Generalised anxiety – behaviours not usually seen in a neurotypical person of the same age may be anxiety – related. Look for rapid breathing, breath holding, worsening hand-wringing. Can also drive seizures and dystonia. Episodic anxiety (panic) – episodes lasting up to 30 minutes where other behaviours/breathing symptoms (and everything else, becomes significantly worse. Appear distressed/terrified. See Figure 2 in Singh and Santosh Emotional Dysregulation.	**All-encompassing approach whereby (I) emotion, (II) behaviour and (III) autonomic function needs to be considered holistically in patients with Rett.** Explore other potential untreated issues, which could be causing pain leading to distress/anxiety, including but not limited to, reflux, gall-bladder, UTI/Urine retention, dental, broken bones/injury. Conventional anxiety treatments (e.g. SSRIs) are often not successful and can lower the seizure threshold.	Helping children with Rett through depression and anxiety. What is meant by Emotional Dysregulation and Behavioural Disorders? By Amy Blake – not published. A. Hryniewiecka-Jaworska et al. Prevalence and associated features of depression in women with Rett Syndrome, E. Foden, M. Kerr, D. Felce, A. Clarke. Singh and Santosh. Key issues in Rett Syndrome: emotional, behavioural and autonomic dysregulation (EBAD) - a target for clinical trials. Orphanet Journal of Rare Diseases. 2018 Jul 31;13(1):128.

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
See also Paroxysmal episodes not due to epilepsy	Mood lability. Loss of interest in normal day to day activites. Children and adults with Rett Syndrome are as vulnerable to typical sadness triggers as general population. Consider current/recent personal circumstances when reviewing EBAD issues. **Autonomic Dysregulation** Breathing difficulties such as apnoea and rapid breathing. Cardiac problems – sudden swings in heart rate (tachycardia/bradycardia). Gastrointestinal problems – constipation and loose stools. Flushing and sweating. **Behavioural Dysregulation** Repetitive rocking, screaming, scratching, agitation, sleep problems.	Benzodiazepines for control of seizures should be used with extreme caution as may lead to respiratory depression and could be life-threatening in individuals with Rett Syndrome. Drugs which may prolong the QT interval such as common antibiotics or other psychiatric medications should be used with caution. Referral to Centre for Interventional Paediatric Pharmacology and Rare Diseases. Explore all possible physical causes for sudden changes in mood or behaviour. Acute medical conditions common in Rett Syndrome include UTIs, urinary retention, gall bladder sludge and stones. Pharmacological interventions in a specialist setting only in case of effects on the autonomic symptoms. Helpful if parents/carers can make video recording of episodes. 48 hour EEG with video telemetry and sleep study should be done at baseline, including neurophysiological, cardiological and respiratory measurements (at minimum) to distinguish from seizures.	

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Mood Problems Including Depression	*Low mood:* **Would the presentation be usual for a neurotypical person of the same age? If not, then the problem should be treated.** Look for loss of interest/enjoyment in previously pleasurable activities, reduced attempts to communicate, increased lethargy, increased distress – assess degree of change from their own baseline and degree of impairment. Establish if the problem is related to poor quality sleep. Refer for sleep study including neurophysiological, cardiological and respiratory measurements to identify any sleep issues which are not being addressed. Can be prolonged in some people or present in some for some of the time. Mood lability common; uncontrolled giggling/laughing spells. Explore all possible physical causes for sudden changes in mood or behaviour. Acute medical conditions common in Rett Syndrome include UTIs, urinary retention, gall bladder sludge and stones.	May also experience worsening seizures, gastrointestinal symptoms, tachycardia, blood pressure, sweating. Conservative management techniques – PBS, music, environmental strategies, distraction, calming/soothing techniques suitable to the individual. Conventional treatments (e.g. SSRIs) need to be used very cautiously and may not be very successful. Benzodiazepines should not be given – this may lead to respiratory depression and could be life-threatening in individuals with Rett Syndrome. Pharmacological interventions in a specialist setting only in case of effects on the autonomic symptoms.	Anxiety-like behaviour in Rett Syndrome: characteristics and assessment by anxiety scales. Prevalence and Associated Features of Depression in Women with Rett Syndrome

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Behavioural Dysregulation Hand stereotypies	**Separated hands:** Tend to improve with time. [Mouthing, hair pulling, pill rolling tremor related to dystonia. One hand behind the neck, castanets, twisting two or three fingers, flapping, tapping, "Sevillana" sequential flexion of metacarpophalangeal and interphalangeal joints 5th to 2nd, hand twirling, hand gaze.] **Joined hands:** Tend to worsen with time and may progress to self-injurious behaviour. [Washing/wringing/clasping, clapping, mouthing]	Refer to OT/physio depending on level of impairment. Look at splinting options – Bamboo splints can be helpful. Neoprene Gloves.	Marisela Dy et al. Defining Hand Stereotypies in Rett Syndrome: A Movement Disorders Perspective (P4.147) Jeffrey Waugh, Nutan Sharma, Heather O'Leary, Kush Kapur, Alissa D'Gama, Mustafa Sahin, David Urion, Walter Kaufmann Neurology Apr 2017, 88 (16 Supplement) P4.147; Hand managment in Rett Syndrome Downs J et al. Perspectives on hand function in girls and women with Rett Syndrome. Dev Neurorehabil. 2014;17(3):210-7. Dy e al. Defining Hand Stereotypies in Rett Syndrome: A Movement Disorders Perspective. Pediatr Neurol. 2017 Oct;75:91-95
Phonic stereotypy Self-stimulatory behaviour, self-injury, agitation	**Other stereotypies:** Arms: Repetitive and rhythmic flexion of the arms. Legs: Intermittent leg elevation and tapping of the floor. - Toe walking, jumping - Feet twirling - Whole body: trunk rocking, shifting weight from one leg to the other - Repetitive sounds - Repetitive words or phrases - Rocking, self-touching - Scratching, biting, mouthing, wringing causing skin breakdown, head banging - Behavioural disturbance associated with distress.		

Renal/Urological

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
UTI/Renal	UTI's vs Urinary Retention – two separate things. Urinary retention can be related to certain meds. Urinary retention can cause repeat UTIs.	Refer to Urology for assessment in case of urethral blockage. Cross check medication re UR side-effect. Urinary retention can be related to certain medications. Cros- check prolonged use of medications which can cause UR e.g. anti-histamines, anticholinergics/antispasmodics, trycyclic antidepressants. Ultrasound to assess urinary retention.	UTI infections – Reducing the Risk (Rett UK Family Companion Factsheet) Bas et al. Report of the first case of precocious puberty in Rett Syndrome, J Pediatr Endocrinol-Metab 2013;26(9-10):937-9 Giesbers et al. Incontinence in Individuals with Rett Syndrome: A Comparative Study, J Dev Phys Disabil.2012 Jun;24(3): 287-300

Gynaecological

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Gynaecological	Polycystic Ovaries Syndrome (POS) often as result of prolonged use of Sodium Valproate.	Annual well woman checks. Cancer screening as per neurotypical population.	

Endocrine

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Puberty	Hormonal changes can worsen all EBAD symptoms and affect seizures and tone. The efficacy of medication can reduce at this time. Likely to have irregular menses but check also for polycystic ovaries. May also be hairier. Growth and nutrition need closer monitoring during adolescense. Screaming, mood disturbances, loss of appetite, period pains. Can worsen tone/epilepsy etc.	For polycystic ovaries – blood tests for high levels of male hormones and scan of the ovaries. Use over the counter painkillers as needed for period pain.	Puberty in Rett Syndrome – Family Companion On Line - Rett UK – by Dr Hilary Cass Killian et al. Pubertal Development in Rett Syndrome Deviates from Typical Females, Pediatr Neurol. 2014 Dec;51(6): 769-775 Puberty Questions and Answers – rettysyndrome.org Knight et al. Pubertal Trajectory in females with Rett Syndrome: a population-based study, Brain Dev. 2013 Nov; 35(10): 912-20 Bas et al. Report of the first case of precocious puberty in Rett Syndrome, J Pediatr Endocrinol-Metab 2013;26(9-10):937-9
SIADH	Increase in seizure activity if epileptic but can cause seizures in someone who previously has not had them. Increased secretions. Visual disturbances. Low sodium in blood tests. Agitation, nausea and tiredness.	Referral to endocrinologist. Sodium supplement. May need restricted fluid regime.	Norsa et al Chronic Hyponatriemia Associated With Rett Syndrome Pediatric Neurology, Volume 50, Issue 1, e1 - e2

Musculoskeletal

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Mobility	Gross motor skills are delayed or sometimes lost later but not in all patients. Gaited walk, toe walking and contractures are common. Maintaining mobility and function is critical to the long term health and well-being of patients with Rett Syndrome. Achievement of all developmental milestones may not always be possible but the ability of some individuals with Rett Syndrome should not be underestimated. All goals should be planned on an individual basis. Some individuals do not achieve independent standing or walking ability. It is important to provide a means of assisting these skills with specialised equipment which is under regular review. Budget provision is made for this purpose. Individuals with Rett struggle with motor planning necessary to change from one position to another. Postural transitioning is as vital as sitting and standing in terms of independence. Regular therapy review and guidance for care givers is vital to develop these skills and/or to maintain the ability to transfer independently for as long as possible. Manual handling policies may also inadvertently deny experience and opportunity to individuals who have achieved these skills, particularly as they get older. This can negatively impact other aspects of physical and emotional health and well-being including; bowel function, sleep/wake cycle, socially, scoliosis and communication.	Refer to wheelchair services and physiotherapy. Patients who are able to, should be encouraged to actively take part in exercise to whatever degree they are able to improve overall prognosis. Individuals with Rett Syndrome who are able to weight bear in standing position, with or without support, should be encouraged to use this skill as much as possible for a minimum of 2 hours per day. All non-ambulatory individuals with Rett Syndrome should have access to specific standing equipment tailored to their needs and use it for a total of at least 2 hours per day. This equipment should be reviewed by a therapy team and serviced on a regular basis with adequate provision of funds for repair and replacement as required. Standing equipment should be used on a daily basis and incorporated into meaningful activity at home or school environment. Care givers should be given guidance and support by the therapy team to achieve this. Orthoses if used should be routinely reviewed. Budget provision should be planned for this and orthoses replaced/altered as the need arises. Active dynamic moment is vital for children and adults with Rett Syndrome. Activities such as water play, horse riding, use of adapted tricycles and daily exercise can help in maintaining physical skills. Patients should be encouraged to actively take part in exercise to whatever degree they are able, to improve overall prognosis.	Larsson et al Normal reactions to orthostatic stress in Rett Syndrome 2013 Jun;34(6):1897-905. doi: 10.1016/j.ridd.2013.02.027. Epub 2013 Apr 11

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Mobility (continuted)	Active preservation of mobility is crucial for every aspect of the individual's welfare that alternative provision is put in place to maintain mobility and function.	Individuals with Rett struggle with motor planning necessary to change from one position to another. Postural transitioning is as vital as sitting and standing in terms of independence. Regular therapy review and guidance for care givers is vital to develop these skills and/or to maintain the ability to transfer independently for as long as possible. Vigilance regarding injury, pressure sores, anxiety and other problems is vital as it will impact on the individual to reach full physical potential. Issues should be addressed immediately. **Pro-active early response and vigilance in trouble-shooting any problem that interferes with mobility is key.**	
Scoliosis	Affects approx. 80% of patients. Asymmetries in movement and range of movement. Higher risk in low muscle tone and non-mobile group.	Consider impact on lung function. Annual checks but bi-annual during puberty. Referral to orthopaedic team. Continual monitoring of spinal integrity. Dynamic bracing. Promote standing and weight bearing minimum 2hrs per day. Risk of curve continuing to increase into adulthood – continue to monitor. Any change – refer back to spinal team.	Scoliosis in Rett Syndrome - A collaboration between parents, clinicians and researchers. Orthopaedics for Kids: Scoliosis in Rett Syndrome.

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Osteopenia Osteoporosis	Risk of fractures – low threshold for X Rays as pain can also be masked. Anti-convulsant medications increase risk especially sodium valproate. Fractures, low bone density.	Vitamin D deficiency is common in children and adults with Rett Syndrome. Blood test to check vitamin D levels at baseline. Six week course of high dose Vit D for adults identified with Vit D deficiency (20,000 iu weekly) before commencing 800iu recommended daily for adults as well as calcium supplements. For children and young people aged 1 yr and older, supplement 400iu daily. Consider Dexascan. Bisphosphonates – tablet or injection. Joint Deformities including leg length discrepancy and hip subluxation/risk of dislocation	Zysman al. Osteoporisis in Rett Syndrome: A Study on Normal ValuesLilit Zysman1, Meir Lotan 1,2*, and Bruria Ben-Zeev1, ScientificWorldJournal. 2006 Dec 15;6:1619-30. Lambert et al. Lower incidence of fracture after IV bisphosphonates in girls with Rett Syndrome and severe bone fragility. PLOS One 2017; 12(10):e0186941 Jefferson et al. Clinical Guidelines for Management of Bone Health in Rett Syndrome Based on Expert Consensus and Available Evidence. PLOS One 2016 Feb 5:11(2):e0146824 Shore et al. The role for hip surveillance in children with cerebral palsy. Curr Rev Musculoskelet Med. 2012 June; 5(2) 126-134
Joint Deformities including leg length discrepancy and hip subluxation/risk of dislocation	Constant visual check. Vigilance around symmetry. Weightbearing. Subtle changes in posture. Recognise change and possible pain from hips which may degenerate in older people.	Encourage functional activity to the individual's potential. MDT/OT/ Orthopaedic Referral. Regular changes of position which aims to maintain a full range of movement through all joints. Maintaining walking and standing for as long as possible. Hips and spine should be checked annually by X Ray. Hip resurfacing may relieve pain. Gabapentin may be of benefit in pain related to hip subluxation, as may focal botulinum toxin injections.	Lotan, M., Merrick, J. (2011). Rett Syndrome: Therapeutic interventions. Nova Science Publishers. Lotan, M. (2006). Rett Syndrome, Guidelines for Individual Intervention. The Scientific World Journal, 6(6), 1504-1516. doi:10.1100/tsw.2006.252 The role for hip surveillance in children with Cerebral Palsy.

Dental

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Dental	Tooth decay. Dental trauma following accidents, seizures and falls. Tooth wear - grinding associated with bruxism. Gum disease - bleeding gums and loose teeth Mouth ulcers. Hypersalivation - increased salivation. Medication related osteonecrosis of the jaw (particularly in relation to bisphosphonate / anti-resorptive medications). Risks associated with treatment which requires sedation or general anaesthesia. Bruxism is a recognized sleep disorder, which should be assessed through full polysomnography as this process causes arousals which disturb sleep leading to low mood.	Refer children and adults to the local Community Dental Service or contact local Dental Hospital Department of Paediatric Dentistry or Special Care. Dentistry. High fluoride toothpaste. Corsodyl gel. Fissure sealants. Dental checks may be necessary under sedation.	How to find a special care dentist BSDH Clinical guidelines Oral care and people with learning disabilities Clinical Guidelines and Integrated Care Pathways for the Oral Health Care of People with Learning Disabilities

Social

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Transition	Planning should start at 14. Introduce to GP – annual (getting to know me) appointment. Early referral to: Community Learning Disability Nursing Team Community Physio Community OT Community SaLT Community Mental Health services as appropriate Referral from paediatric consultant care to adult teams. NHS Continuing Health Care as appropriate.	Neuro rehabilitation medicine to take responsibility.	Transition to Adulthood Learning Disability Health Check Toolkit

Glossary

ARDs	Anti Reflux Drugs
BSPD	British Society of Paediatric Dentistry
BSDH	British Society of Dental Health
BMI	Body Mass Index
CT Scan	Computerised tomography scan uses X-rays and a computer to create detailed images of the inside of the body
EBAD	Emotional, Behavioural and Autonomic Dysregulation
ECG	How heart beats are measured
EEG	An electroencephalogram is a test used to find problems related to electrical activity of the brain
GI	Gastrointestinal
MRI	Magnetic Resonance Imaging is a type of scan that uses strong magnetic fields and radio waves to produce detailed images of the inside of the body
MDA	Multi-Disciplinary Assessment
NG	Tube – Naso gastric tube
OT	Occupational Therapy
PPI	Proton pump inhibitors
PBS	Positive Behaviour Strategies
PNS	Parasympathetic Nervous System
QT Interval	The QT interval is the time between two points in each heart beat
RTT	Rett Syndrome
SSRIs	Selective Serotonin Reuptake Inhibitors
SNS	Sympathetic Nervous System
SIADH	Syndrome of inappropriate antidiuretic hormone secretion is a condition in which the body makes too much antidiuretic hormone (ADH)
UTI	Urinary Tract Infection

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Notes

On behalf of all the children and adults with Rett Syndrome and related disorders, our special thanks to the Bolland Family Charitable Fund for their kind support for the production of the Rett Syndrome Health Checklist