

RETT UK · REVERSE RETT · FOXG1 UK

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Consultant Paediatric Neurologist

# Patient Organisations/Representatives

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Hilary Truss

The Rett Syndrome Health Checklist was compiled and edited as a collaborative piece of work by Becky Jenner. Rachael Stevenson and designed by Wesley Havill.

# Introduction

#### What is Rett syndrome?

Rest Syndrome is a rare neurological disorder affecting mainly females and very few males. It is present from conception and usually remains undetected and feeding-permited delay occurs sound one year of age, when children may lose exquired skills and become withdrawn. It should be noted though that some children may regress later and/or have minimal regression. Rest Syndrome is usually cusued by a fault (mutation) on a gene called MECPy which is found on the xill becomes one. In 1998 of case it is a new mutation and not inherited. There are many different types of mutation and not inherited. There are many different pyes of mutation and not inherited. There are many different pyes of mutation and not inherited. There are many different pyes of mutation and not inherited.

#### 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, obtain kins for further reading including relevant research oasers.

#### 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 asknowledgements regarding the professionals who have been involved and their disciplines.

Rett Disorders Alliance UK Rett Syndrome Health Checklis

### 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 at 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 Ret Syndrome does not exclude the person from developing other problems that you would see in the neuropipical population, but syndrome range he maked by the completely of the disability and the persons invaled bally to communicate. Taking and isteining to parents and cares who know the person neally well is key to understanding the nature of the problem and what is typical/abypical' 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.

Rett Disorders Alliance UK Rett Syndrome Health Checklist

### Stages of Rett Syndrome

Red Syndrome has been described as being in four stages: early signs, regression, plateau and late motor deterioration (Refer to NHS Choices Red Syndrome). This can be applied in general terms but there is a spectrum in severy and environmental lations can efficience for 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 regulates at the severe end of the spectrum regulates at the severe described in the severe end of the spectrum regulates at the same diseases profession engineer dealfer on. Some constitute to a severe described the severe end of the spectrum regulates at the same diseases profession engineer dealfer on. Some constitute to a severe deal of the spectrum of

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 instituted as they would be for a neurotroical person presenting at A & E or as an impatient.

# Rett 'episodes'

Some Rett Iterature, 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 school the made to next read the seess and informace the nonblem.

# 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 paediamician up to age 18, it is very important that the GP does have some knowledge and experience of the young pesson 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.

Rett Disorders Alliance UK Rett Syndrome Health Checklist

#### 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 gaz 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 shifty which may also be inflammed by which environment to some extent, but the important message here a root to assume they do not understant. Find out how they communicate, give them every opportunity to be included in conversations, give them the opportunity to express their opinions and windows. In other words,

Referencies:
Culcision T, Leliber C, DxGregorio G, Vogel-Furley V, Bames K, Kaufmann WE, et al. Adapting the Multien Scales of Early Learning for a standardized measure of development in children with Rest syndroms. Intellectual and Developmental Disabilisies. 2017;55(9):4193. Notorisia-Acea J, Polick O, Sauf E, Wine L, Silberg T, Nosenkom A, et al. Assessing cognitive functioning in females with this trayintome by eye-acting methodology. Europea-bound of Praediction Notorings 2018;2(1):34–45.

## Key Principles For Parents

You know your sondaughter 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 Rest UK Health Passoor of the Reverse Ret Heosital Passoort.

The Health-Heighal Prosports will be updated regularly so please refer to Rest LV or Revesse Reti websites to check for updated information. Many lamilles find it really heighal to connect with other families through social media (where are two groups in the UK. Rest Syndrome or heigh social s

### Advanced care planning

Having an advance 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, to recample, the nurses, doctors, care home manager or family members. Depending on the capacity of the individual with Rest Syndrome, they may observe to express some views, preferences and wides about their future care. Where this is possible, every reasonable effort should be made to allow the preference of the care of the care of the care. Where this is possible, every reasonable effort should be made to allow their family and people who know them very well that will be mading 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 the children with Rest Syndrome are diagnosed much quicker and earlier than they are at the moment. A world where we known than the control and the condition and of the condition and the factors that affect that progression. A world where are very stage of development, the tenter are available to mitigate against the distressing symptoms which can occur and, utilinately, a world where treatments are available on which can be considered and allow accorded with Rest Syndrome to be no morall with care the condition and allow accorded with Rest Syndrome to be no morall with a care to accord to the syndrome of the condition and allow accorded with Rest Syndrome to be no morall with a care to accord to the syndrome of the syndrome of

www.reverserett.org.uk



#### 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 Fett 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.

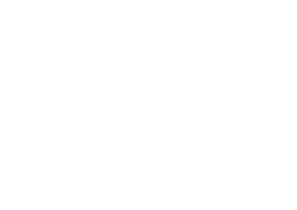
As FOXG1 Syndrome is a relatively newly identified genetic condition, there are no long term studies into the health or longevity of people diagnosed with the syndrome. For a few children it is clearly a life-limiting condition but there is every reason to believe that the majority will go on to live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

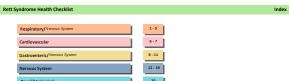
Clinicians who have knowledge of FOXG1 Syndrome are as rare as the syndrome itself, so families affected by this condition will also benefit from having access to the Rett Disorders Health Checklist

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

www.foxg1.uk

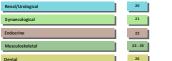
# Rett Syndrome Health Checklist





27

29



Glossary

Index



feeding

 Frequent spitting up or vomiting Recurring pneumonia or respiratory infections Less than normal weight gain or growth

(The arridantal inhalation of food fluid or reflex into the

Aspiration

Arching or stiffening of the body during - Irritability or lack of alertness during feeding Failure to accept different textures of food Long feeding times

What to look forthe aware of

Difficulty chewing Difficulty swallowing Difficulty breast feeding in infants Coughing or gagging during meals

Excessive drooling or food/liquid coming out Difficulty co-ordinating breathing with eating and drinking

Increased stuffiness during meals Guroly, hoarse or breathy voice quality

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

Investigation & treatment and/or referral pathway

Video-fluroscopy/barium swallow if aspiration or unsafe

swallow is suspected. Check for reflux symptoms and address actively.

Inform carers of precautionary steps: meals/eating/drinking/tube feeding and for 30 mins after.

Rett Syndrome Hea	alth Checklist		Respiratory/ Nervous System
Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Silent aspiration	White over secretarion can cause subten on conclusive symptoms, in stear aspiration, there is no coughing or chearing.  - Real watery eyes  - Colour changes to skin around the eyes  - Chroning so be a cought of the eyes  - Chroning so bearing voice  - Splayed hands in younger children		
Chest infections   Preventative	Prevention of check infections is key to the languisty of patients with flax.  Early management of active infections leads to better outcomes for this patient population.  Regular check infections, swallowing	Annual flu veccination for patient and carers.  PPV/Promissions for patients over the age of 2 because of you give integration, because of you for the patients of your fluor control of the control of t	Citis et 2 Subsequentia et cara la Apprilian Dissammen in Nego - nal Rossa Model for Ret Systemes Sci Rep. 2027/(1) 2022. Published 2027 Sep 20. des 10.1038/s41598-017-12293-8
strategies	difficulties, aspiration. Increased secretions. If coughing becomes unusually frequent, observe carefully and take temperature regularly.	be useful.  Daily mucodyne (carbocysteine) to keep secretions more fluid and eatier in cough up.  If temperature is raised, the patient appears unwell and or secretions are difficult to clear, notify GP and chest physio immediately.	
			Respiratory/ Nervous System 3

-,			
<ul> <li>Managing active infections</li> </ul>	Raised temperature. Increased secretions.	If chest infection is confirmed, stop prophylactic anti- biotics if in place and start rescue anti-biotics immediately.	Don't leave it to chance article -Yvonne Milne MBE
	Generally unwell.	Increase dose of Mucodyne as appropriate.	
	Emergency anti-biotics specifically for active chest infections can be stored at home for immediate use in line with the individual patient's specified	Use of nebuliser for saline or salbutamol inhalation as needed.	
	respiratory protocol, in order to avoid inevitable delays.	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).	

Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Cardiovascular dysregulation	Oyengulation of cardiovascular parameters. Heart size, Bir peripheral vascoonstriction, may change markedy within seconds.	Monitor for QTC protongation at least annually. QTC may vary markedly within a short space of time. Bewere old avoid medications causing QTC protongation.	Jaiu & Dispertroms, Assessment of the manufactured registrations, and the histogeneous phenotypes and tacilities the indices review to the histogeneous phenotypes and tacilities continuous and of that systems, the continuous continuous and that systems are also as the continuous and the continuous

Rett Syndrome Health Checklist

# Kumar et al. Cardiovescular autonomic dysfunction in children and adolescents with Rett Syndrome. Pediatr Neurol. 2017 May;70:61-66. 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.

Cardiovascular

Cardiovascular 7

Rett Syndrome He	ealth Checklist		Gastroenteric
Symptoms	What to look foribe aware of	Investigation & treatment and/or referral pathway	Further Reading
Growth and nutrition	Poor weeky gain, yolinged feeding insen, poor find rottee, chrisen god molecular gifficular chrisen gain on the chrisen gain of the chrisen gain gain gain gain gain gain gain gai	singst, weight and BM foliable the monotoned regularly. Counter Floration for Manufaction Regularities of proposal with the Structure and a soliable floration of proposal with the Structure are available from Floration Flor	exposing both accurace in the synchronic introduction and Depoles have, in climations booked for behalf and comes booked for behalf and comes and and call Commission Placement regression and and call commission of the commission synchronic Journal of Productic Commission synchronic Journal of Productic Commission and Technology 2017, 242 and Technology 2017, 243 and Tec
Gastroenterology	Officialize include reflux, dearnous (SMS) overactively, consistent personal results of solution (PMS overactively) associated pain, ventiling, agmostly), associated pain, ventiling, agmostly, water for approximate production of the property of the ARDS. Failure to empty bowels completely especially in other people. Impacted bowels – diarnhoea may be sign.	Movest, Lauritanis, appropriations may hely eith conscipation as well are natural remedies such as prune jack.  Use ARDS guidelines should this occur.	Intels Leaved Caston Associated Studies in Red syndrome Chandle for derivation on issessment and management. As the control of the control of the Model et al. Castoniamy Placement Emprove Height and Whatful Castoniamy Placement Emprove Height and Whatful Castoniams of the Syndrome, Journal 207-207 207 207 207 207 207 207 207 207 207

Rett Syndrome Health Checklist			Gastroenteric
Symptoms	What to look for be aware of	Investigation & treatment and/or referral pathway	Further Reading
Symptoms  Eating and derinking officialities Annous System	What to both forthe aware or promises Feeding when the promises I have been coughing engine. I he may be set, coughing, peopling.	Investigation & Invasiment audior referral painway Feeding and binduscus attempts, must, throughest and or thickness feed backs.  Control good and the state of t	Further fleading
			Gastroenteric 9

Rett Syndrome He	ealth Checklist		Gastroenteric
Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Califibladder     Gallibladder     Parior ealitis	Paintiblecomfort but can be mailed. Feer, junction and ventiling. Committee, surprise and ventiling. Committee, surprise vice the paintible vice of the paintible vice vice vice vice vice vice vice vic	Check for choiceyattis, gallitarios and gallitacher skrige.  After exchange genocouphager etnic, gallitacher skrige.  After exchange genocouphager etnic, gallitacher skriger and proposed gallitacher skriger.  Bernard et al. (1997) and skriger etnic skriger etnic skriger etnic skriger etnic skriger etnic skriger.  Physical examination, Milk CT scan and ulmonound.	The Register of Annaectic Control Annaectic Control
			Gastroenteric 11

Neul et al. Rett Syndrome: Revised diagnostic criteria

Neul et al. Developmental delay in Rett Syndrome:

Data from the natural history study. J Neurodey.

Disord, 2014; 6 (1); 20 Marschik et al. Early development in Rett Syndromethe benefits and difficulties of a birth cohort approach. Developmental Neurorehabilitation 2018: 21: 68-72

and nomenclature. Annels of Neurology 2010: 68: 944-

seen in early

Muscle Tone and

Tone Management

have a clear period of regression where skills are

Variable from low to high tone but frequently

outside the normal range. See also autonomic dysfunction - hypertonia.

What to look for the aware of

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.

Holistically assess with MDT for neurological disorders.

Check positioning - standing (frame), appropriate supported seating, lying (sleep system). Check for co-morbidities (constination) and medications

Investigation & treatment and/or referral nathway

Referral to appropriate services.

Lotan, M. (2006). Rett Syndrome, Guidelines for Individual Intervention. The Scientific World Journal,

Nervous System

Typical medication used for managing spasticity: bacinfen

Rett Syndrome Health Checklist

		Inny can aso be pamui. See aso autonomic dysfunction - hypertonia		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.	Kadyan V et al. Intrathecal bacicfien for spasticity management in Rett syndrome. Am J Phys Med Rehabil. 2003;82(7):560-2.
				Tomodo T et al

Bodularium train can be considered for focal elements of high trore (letther specificity or dystatosis).

Whenever discussion in the syndrome are analysis of 00 papers with described MEXTS mutation and 100 papers with described with

Altered Tissue
Properties

Over the an any abnormal modes contraction con
plant in department of the contraction of the contrac

measure of existance to control, electrolly set produces a visitant to organize happened project sendors you without to organize happened project for the control of the control of the control of the control of the fixed muccularised and offermity.

Nervous System

Humphreys P. Barrowman N. The Incidence and Evolution

**Eurther Reading** 

Mood Jability

What to look for he aware of

Breathing difficulties such as annoga and rapid

stools. Flushing and sweating.

Cardiac problems - sudden swinns in heart rate Gastrointestinal problems - constination and loose

Behavioural Dysregulation

Repetitive rocking, screaming, scratching, agitation, sleep problems.

Investigation & treatment and/or referral nathway

with extreme caution as may lead to respiratory decression

antibiotics or other osvehiatric medications should be used

Explore all possible physical causes for sudden changes in mood or behaviour. Acute medical conditions common in Rett Syndrome include LITIs urinary retention, nall bladder sludge and stones.

48 hour EEG with video telemetry and sleen study should

Phonic stereotypy

hebaviour self-injury

anitation

Other stereotypies: - Feet twirling leg to the other - Repetitive sounds

> Rocking, self-touching - Scratching, biting, mouthing, wringing causing skin breakdown, head banging Behavioural disturbance associated with distress.

Rett Syndrome Health Checklist

Arms: Repetitive and rhythmic flexion of the arms. Legs: Intermittent leg elevation and tapping of the floor. Toe walking, jumping - Whole body: trunk rocking, shifting weight from - Repetitive words or phrases

Dy e al. Defining Hand Stereotypies in Rett Syndrome

Nervous System

Rett Syndrom	Health Checklist		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.	Refer to Urology for assessment in case of urethral blockage.	UTI infections – Reducing the Risk (Rett UK Family Companion Factsheet)
	Urinary retention can cause repeat UTs.	Cross check medication re U R side effect.  Ulrinary retention can be related to certain medications.  Cross check prolonged use of medications which can cause  Lift e.g. and histamines, articolaterajcis/artispassmodics,  Psycycle artificipations.  Ultrasound to assess ulrinary retention.	Base et al. Region of the first case of processors publishy in Rise Uniformes, 2 Produce Endocrisci- Metale 2012;76(9-17):937-9 Geoderns et al. Inconfirmence in Individuals with Rise Synthesis: A Comparative Statey, 2 Dev Phys Deater, 2012;7 Jun;24(3): 287-303

Rett Syndrome Health Checklist Gynaecologics			Gynaecological
Sympto	s What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
<ul> <li>Gynaecologic</li> </ul>	Polycystic Ovaries Syndrome (POS) often as result of prolonged use of Sodium Valproate.	Annual well woman checks.  Cancer screening as per neurotypical population.	
	•	I	

Rett Syndrome	Health Checklist		Endocrine
Symptoms	What to look for/be aware of	Investigation & treatment and/or referral pathway	Further Reading
Puberty	Hommand Language can worsen and ERAD opprehension of their solories and home motions of their Stimms, and their solories and th	For pulsage of course. Hood lasts for high levels of neish homeous and use of the outles.  Use out the counter particulars as needed for period pan.	Noting in Net systems – Family Companies Christin- Red Ut-1-(1994) Cases Vision et al. Publical Davidgement in Red Spiriture Schools (1994) Cases Paulin Teach (1994) Cases Paulin Teacher
SIADH	Increase in subserve activity of enterptic bot can cause solutions in common with graveloody has not had from.  Visual disarrhores. Low societies in the disarrhores. Application, reasses and treatheres.	Referred to endocronologist. Sodium supplement. May need reserches thair regime.	States at Common Injuriorations Associated IRIS Mark Systems Pediatric Numering, Volume SC, Nase 1, 41 - 42
			Endocrine 22

# Mobility

S What to look for/be aware of Inve Gross motor skills are delayed or sometimes lost later but not in all patients.

Gaited walk, toe walking and contractures are

Maintaining mobility and function is critical to the long term health and well-being of patients with Rett.

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

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 olivers is vital to develop these skills and/or to

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

Manual handling policies may also inadvertiently 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 scollosis and communication.

## Refer to wheelchair services and physiotherapy. Larse

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 still 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 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 disty basis and incorporated into meaningful activity at home or school environment. Care others should be used to when outdance and support by the therapy

Orthoses if used should be routinely reviewed. Budget provision should be planned for this and orthoses replaced/altered as the need arises.

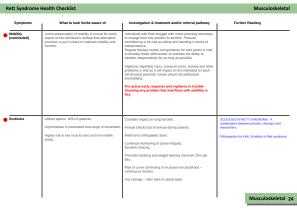
able, to improve overall prognosis.

team to achieve this.

Active dynamic moment is vital for children and adults with Rest 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 de

# Further Reading Larsson et al Normal reactions to orthostatic stress in Rett syndrome 2013 Jun;34(6):1897-905. doi:

1016lj ridd 2013.02.027. Epub 2013 Apr 11



Six week course of high dose Vit D for adults identified with

Rett Syndrome Health Checklist

Vit D deficiency (20 000 is weekly) before commencion 800iu recommended daily for adults as well as calcium 12(10):e0186941 For children and young people aged 1 yr and older, tefferons et al. Clinical Cuidolines for Based on Expert Consensus and Available Risphosphonates - tablet or injection

Joint Deformities Lotan, M., Merrick, J. (2011). Rett syndrome: including leg Therapeutic interventions. Nova Science length discrepancy and hin subluvation/risk of Lotton M. (2006). But Sundanno, Cuidalines for dislocation range of movement through all joints. Maintaining walking The role for hip surveillance in children with cerebral palsy

Musculoskolotal

Lambort et al. Lower incidence of fracture after

Tooth decay.

falls.

What to look foribe aware of

Dental trauma following accidents, seizures and

Tooth wear - orinding associated with bruxism.

Gum disease - bleeding gums and loose teeth

Medication related osteonecrosis of the law

(particularly in relation to bisphosphonate / antiresorptive medications). Risks associated with treatment which requires sedation or general anaesthesia. Bruxism is a recognized sleep disorder, which should be assessed through full polysomnography

Hypersalivation - increased salivation.

Further Reading

Oral care and people with learning disabilities

Clinical Guidelines and Integrated Care Pathways for

the Oral Health Care of People with Learning

**BSDH Clinical guidelines** 

Disabilities

as this process causes arousals which disturb sleep leading to low mood.	

Investigation & treatment and/or referral pathway

Refer children and adults to the local Community Dental

Service or contact local Dental Hospital Department of

Dental checks may be necessary under sedation.

Paediatric Dentistry or Special Care.

High fluoride toothpaste.

Dentistry.

Corsodyl gel.

Fissure sealants.



#### Social Rett Syndrome Health Checklist Symptoms What to look for/be aware of Investigation & treatment and/or referral pathway Further Reading Transition Planning should start at 14. Neum rehabilitation medicine to take Transition to Adulthood Learning Disability Health Check Toolkit Introduce to GP - annual (getting to know me)

Community Learning Disability Nursing Team Community SaLT Community Mental Health services as appropriate Referral from paediatric consultant care to adult NHS Continuing Health Care as appropriate.

ARD's Anti Reflux Drugs
RSPD Rritish Society of Paediatric Dentish

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 Mannets. Resonance limating is a type of scan that uses strong magnetic fields and radio waves to produce detailed images of the inside of the body

MDT Multi-Disciplinary Assessm
NG Tube – Naso gastric tube
OT Occupational Therapy

PI Proton pump inhibitors BS Positive Behaviour Strategies NS Parasymoathetic Nervous System

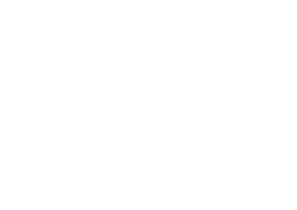
QT Interval The QT interval is the time between two points in each heart beat

SSRI's Selective Serotonin Reuptake Inhibitors

Sympathetic Nervous System Syndrome of Inappropriate antidiuretic hormone secretion is a condition in which the body makes too much antidiuretic hormone (ADH) Ulriary Tract Infection

Glossary 28

Agitation, 19	Leg length discrepancies, 25
Altered tissue properties, 13	Long OT, 6, 17
Aspiration, 2	Mobility, 23, 24
Autonomic dysregulation, 1, 17	Mood problems, including depression, 18
Breath holding/apnoeas, 1, 17	Movement disorder and motor planning, 15
Breathing difficulties, 1	Muscle tone and management, 12
Cardiovascular dysregulation, 7	Osteopenia, 25
Chest infections, 3	Osteoporosis, 25
Circulation, 7	Pancreatitis, 11
constipation, 8	Paroxysmal episodes (not due to epilepsy), 15
Dental management and issues, 26	Phonic stereotypy, 19
Diarrhoea, 8	Pneumonia, 3
Dystonia, 13	Polycystic Ovaries Syndrome, 21
Early developmental impairment, 12	Puberty, 22
Eating and drinking difficulties, 9	Reflux, 8, 10
EBAD, (Emotional, Behavioural and A	utonomic Scoliosis, 24
Dysregulation), 16	Seizures, 14
Epilepsy, 14	Self-injury, 19
Gallbladder dysfunction, 11	Self-stimulatory behaviour, 19
Gastroenterology, 8	SIADH, 22
Growth and nutrition, 8	Silent aspiration
Gynaecological, 21	Sleep disturbances, 5
Hand stereotypies, 19	Spasticity, 13
Heart rate variation, 7, 17	Sudden change in mood or behaviour, 18
Hip subluxation, risk of dislocation, 25	Transition, 27
Hyperventilation, 1.17	Uncontrolled laughing, 18
Impaction, 8	Urinary retention, 20
Joint deformities, 25	Urinary tract infections, 20



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 and for the production of the Rett Syndrome Health Checklist