

Evelina London Rett Service

This leaflet explains about the specialised multidisciplinary Rett clinic held at the Evelina London Children's Hospital.

About Rett syndrome

Rett syndrome is a profoundly disabling neurological disorder affecting mental processing, including speech and hand use, and brain stem control of posture, movement and cardio-respiratory regulation. The condition is caused by a mutation in the MECP2 gene on the X chromosome. It occurs in 1 in 12,000 girls, whilst boys with the disorder are very rare.

After a period of apparently normal development during the first year or two of life, affected girls suffer a regression in hand skills, gross motor skills, communication and feeding, along with development of the characteristic hand wringing movements.

Those affected by the condition will need lifelong support and care. The bulk of this care is provided by local child development teams, but because any one district will have only a handful of affected children, a team with expertise gained through seeing large numbers of girls with the condition can provide supplementary help and advice.



About the Evelina London Rett Service

The Evelina London Rett Service is a national specialist outpatient service which was established by Dr Hilary Cass in 1992. The service has been developed jointly with Rett UK, demonstrating the benefits of integrated NHS and voluntary sector collaboration. The clinic was initially based at Harper House Children's Service in Hertfordshire, then moved to Great Ormond Street Hospital, and with the support of the Rett UK has also operated as an outreach and teaching service in child development centres around the UK.

In 2009, with Dr Cass' move to Guy's and St Thomas' NHS Foundation Trust, the service has now been re-established at Evelina London Children's Hospital. This has afforded new benefits as a result of involvement from other members of the paediatric neurosciences team at Evelina, as well as the potential for better transition management to adult services.

Although many aspects of the management of girls with Rett syndrome are similar to those with other profound and multiple disabilities, there are some important differences. For example, lack of awareness of the early warning signs in children within the poorer prognostic groups can result in more severe disability and a failure to avoid secondary complications. In addition, since the discovery of the MECP2 gene, we are seeing increasing numbers of girls with a wider range

of disabilities, and are making new discoveries about the clinical course of the disorder. This means that different treatment approaches may be needed.

Over the last 15 years, there have been a number of important roles and outputs from the clinic:

- Approximately 350 patients have been seen, and there is currently an ongoing demand from parents, referrers & Rett UK.
- In addition to running the on-site service, the team has continued to provide a telephone advisory service to clinicians around the UK.
- Members of the clinical team act in a professional advisory capacity to Rett UK providing material for the Rett UK website, talking at regional family events and supporting the Rett UK staff in their work with families.
- There have been a number of papers produced by the clinic addressing a wide range of issues regarding Rett syndrome, such as natural history of the disorder, management of feeding problems, and behavioural characteristics and genetics.
- The team has close working relationships with other interested clinicians across the UK, as well as internationally with parallel national teams in Sweden, Australia, and America.
- Members of the team have run dedicated courses on Rett syndrome and have been invited speakers at national and international conferences.
- The team has collaborated in international guideline development.

The clinical team



Dr Hilary Cass is a consultant in paediatric neurodisability. Her clinical interests include children with autistic spectrum disorders, and management of complex disability with particular reference to feeding and communication problems, and management of patients with Rett syndrome. She is clinical advisor to Rett UK and a number of other voluntary bodies. In addition to publishing widely in the field, she has been an invited speaker at many national and international conferences, including in Australia and New Zealand.

Georgina Feint is a speech and language therapist. Her clinical interests include the management of eating/drinking difficulties, drooling and communication in children with complex disabilities or complex medical problems. She has extensive clinical experience in physical disability, learning disability and complex medical disorders and is the team lead for the speech therapy department at Evelina London Children's Hospital. Georgina is involved with the training of speech and language therapists and mentoring specialist therapists.





Erin Morton is a physiotherapist. Her clinical interests include children with complex movement disorders and orthopaedic/spinal management of children with cerebral palsy and similar conditions. She has clinical experience in assessing and treating children both in acute, outpatient and community settings. She has an extended role with the orthopaedic, spinal and complex movement disorder services within the Evelina Children's Hospital.



Nikki Queton is an occupational therapist. Her clinical interests include children with complex disabilities, upper limb difficulties associated with cerebral palsy and paediatric neurodisability. She has extensive experience in physical disability, learning disability and complex medical disorders both within a hospital setting as well as in the community. She is also involved with children admitted to the neurology ward at the Evelina London Children's Hospital.

About Rett UK

Rett UK is a national charity seeking to make a difference for people living with Rett syndrome. It provides help, advice and support to parents, carers, siblings and professionals – in fact anybody involved with a child or adult who has Rett syndrome.

The aims of Rett UK are to:

Deliver excellent support services that meet the long term needs of the Rett community, reducing isolation and exclusion and improving quality of life, independence, confidence, health and well-being.

Partner with relevant organisations and professionals to fund, raise awareness, resource and deliver key services, projects and campaigns, ensuring that identified Rett community needs are achieved.

Enable the organisation to effectively grow and develop, for the long term benefit of the Rett community.

The services provided include:

- Telephone and electronic support, through a dedicated helpline and email provision, linking families directly to professional support and advice.
- Self-help support groups, located across the UK.
- Rett UK Best Practice Management & Care Guidelines, providing information for medical practitioners and therapists.
- Family Companion – giving information to families about Rett syndrome and other useful information, in an easy to read format.
- A wide range of high quality, up-to-date written information.
- Rett News – a quarterly magazine.
- Regional family events, taking the professionals to local regions and providing information, advice and support.
- Training for health visitors, GP's, social workers and others.

In partnership with the NHS, Rett UK have provided support at clinics since 1992. This support is both practical and emotional. Where there are areas of concern identified, Rett UK supports

families and local delivery teams to provide appropriate help. Often families are faced with difficult decisions and the prospects that their daughters' prognosis may not be as they expect. The Rett UK team are trained to deal with providing the emotional support needed at this time.

The team at Rett UK

Julie Benson was appointed family support manager in July 2012 with over 15 years' experience of working within non-for profit organisation. Julie worked for an advocacy charity providing support to a wide range of adults within the health & social care field. Prior to this she worked for six years in a special school as a teaching assistant. She was working with children with profound and multiple learning disabilities across all ages. Julie is carer to a family member with cerebral palsy and complex needs; this means she has a good insight into the frustration and anguish faced by many families trying to get the best for the person for whom they provide care.



Patrice Hosier was appointed family support officer and is the newest member of the team. Patrice joined Rett UK in November 2012. Previously a registered domiciliary care manager she has experience of supporting residents with Rett syndrome and other complex needs. Her qualifications are extensive all acquired through hard work and dedication. Patrice is responsible for answering the support helpline, responding to queries by email and on Facebook. Patrice organises our regional day and booking speakers.

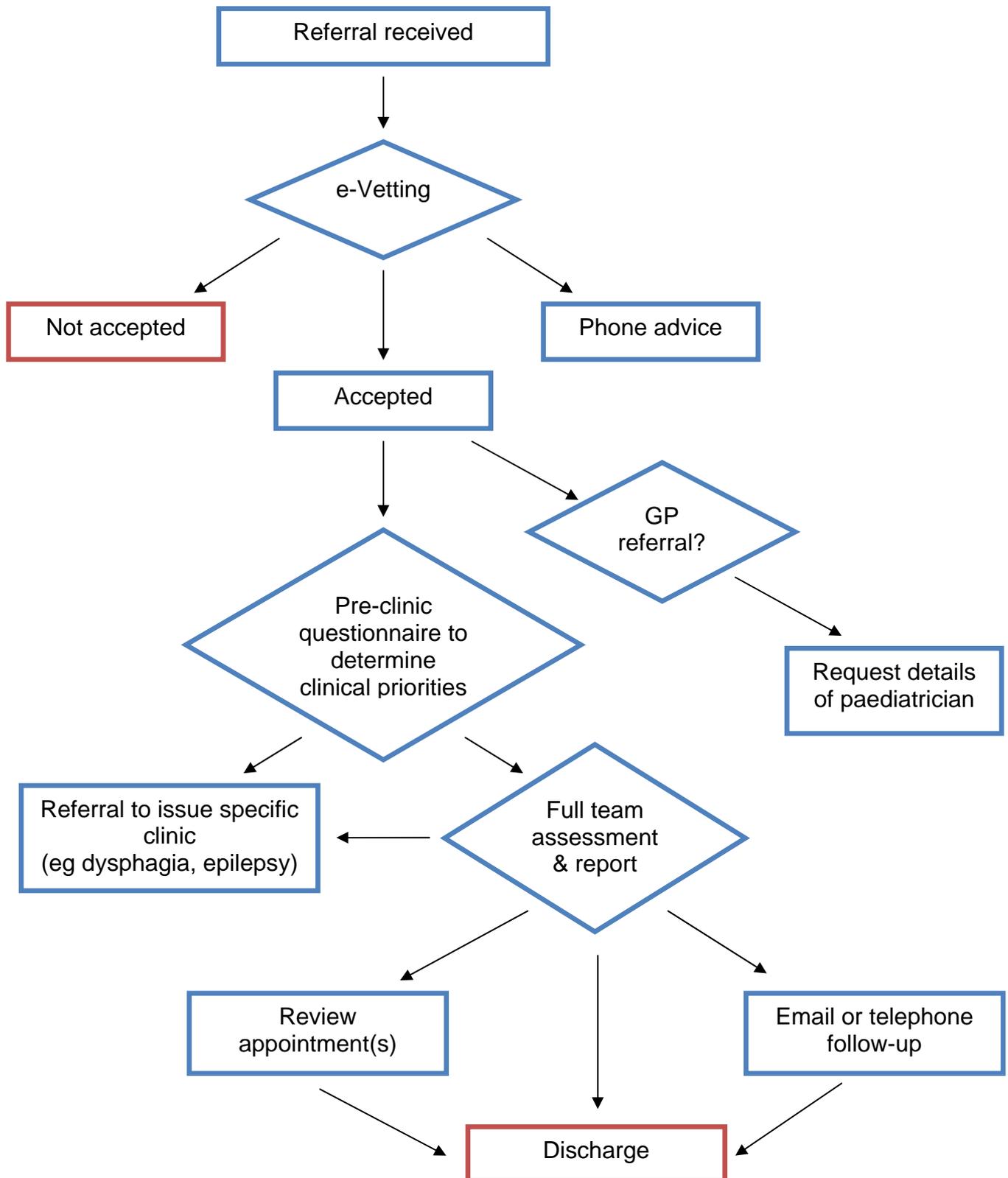
Who can be referred to the clinic?

The team can accept referrals of any patient with genetically confirmed Rett syndrome or clinical features suggestive of Rett syndrome up to the age of 16. Specific indications for referral are as follows:

- Assessment and advice on the movement disorder and/or paroxysmal events, many of which are not epileptic in origin.
- Advice and/or management of the breathing disorder.
- Management of screaming episodes and sleep disturbance.
- Advice on communication and feeding.
- Advice on complex seating issues, as well as OT support for splinting and optimising hand use.
- Scoliosis advice and surgery as appropriate.
- Advice on broader aspects of motor management.
- Assessment and advice on patients not demonstrating the typical mutations and / or undergoing an unusual disease progression.
- General advice on prognosis or the broader management of the condition.

Ideally referrals should come from the secondary level paediatrician, since he or she will continue to lead care locally, but we will also accept GP referrals. Where we receive a GP referral, we will seek permission to contact the child's paediatrician in order to obtain full information on previous assessments and ongoing treatment, as well as to liaise regarding any recommendations that may be made by the Rett service team.

Referral pathway



Initial referral management

- All referrals will be scanned to e-vetting by the Rett clinic secretary.
- Dr Cass will assess whether the referral should be accepted, rejected or managed through phone advice to the referrer.
- Accepted patients will be sent a pre-clinic questionnaire and added to the clinic waiting list.
- For GP referrals, the GP will be asked for contact details of the patient's paediatrician. The paediatrician will be sent a letter informing him/her that the child has been referred to the Rett clinic, and requesting any additional information (except in the unusual situation whereby the GP indicates there has been a breakdown in relationship with the local paediatrician).
- Dr Cass will review the returned questionnaire, and request any additional information needed in advance of the clinic. In some instances, the questionnaire may suggest that it would be better for the child to be seen in a more specific clinic in the first instance, in which case Dr Cass will make a cross referral, with copy letters to the parents and referrer explaining the rationale.

Full team assessment

- The Rett Clinic will run on the 2nd Monday afternoon of the month, with 2–3 children seen in each clinic.
- We will try to see all patients within 18 weeks from referral.
- Based on previous experience, a once per monthly clinic will match the referral rate. In the event that the patient flow exceeds available slots, a clinic will be run on the 4th Monday afternoon of the month, in place of Dr Cass' multidisciplinary team slot.
- The full assessment team will comprise a paediatrician, speech and language therapist with expertise in dysphagia and complex communication impairments, occupational therapist and physiotherapist. Rett UK will also provide a family support worker for the clinic.
- A multidisciplinary report using a standard Rett Clinic template will be completed and sent out within 4 weeks of the appointment to the parents, GP, paediatrician and any other involved professionals requested by the parents.

Follow up

- The majority of children will be discharged after the assessment, with recommendations back to the local team.
- A smaller number of patients may be referred to another clinic (for example epilepsy, botox, movement disorder, sleep) and/or need ongoing email or telephone follow up to the family or referrer to resolve a specific issue (for example management of breathing problems, epilepsy, feeding management and so on).
- A very small minority of patients who have the most complex problems (for example combination of severe epilepsy, dystonia and breathing problems) may need longer term follow up by the full team or one or two members of the team. Past experience demonstrates that this applies to a maximum of 3–4 per year, most commonly with severe epilepsy, who are seen jointly by Dr Cass and Dr Hughes.

Useful sources of information

Rett UK website www.rettuk.org/rettuk-public/rettuk.html

Contact us

If you have any questions or concerns, please contact Layla Georgieu on 020 7188 3998 (Tuesday and Wednesday, 9am to 5pm).

For more information leaflets on conditions, procedures, treatments and services offered at our hospitals, please visit www.guysandstthomas.nhs.uk/leaflets

Evelina London Medicines Helpline

If you have any questions or concerns about your child's medicines, please speak to the staff caring for them or contact our helpline.

t: 020 7188 3003 10am to 5pm, Monday to Friday

e: letstalkmedicines@gstt.nhs.uk

Patient Advice and Liaison Service (PALS)

To make comments or raise concerns about the Trust's services, please contact PALS. Ask a member of staff to direct you to the PALS office or:

t: 020 7188 8801 at St Thomas' **t:** 020 7188 8803 at Guy's **e:** pals@gstt.nhs.uk

Language Support Services

If you need an interpreter or information about your care in a different language or format, please get in touch using the following contact details.

t: 020 7188 8815 **fax:** 020 7188 5953

NHS 111

Offers medical help and advice from fully trained advisers supported by experienced nurses and paramedics. Available over the phone 24 hours a day.

t: 111

NHS Choices

Provides online information and guidance on all aspects of health and healthcare, to help you make choices about your health.

w: www.nhs.uk

Become a member of your local hospitals, and help shape our future

Membership is free and it is completely up to you how much you get involved. To become a member of our Foundation Trust, you need to be 18 years of age or over, live in Lambeth, Southwark, Lewisham, Wandsworth or Westminster or have been a patient at either hospital in the last five years.

To join, please call 0848 143 4017, email members@gstt.nhs.uk or visit www.guysandstthomas.nhs.uk

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