FABRY
RECORD BOOKLET

This booklet is intended for people with Fabry disease.

Introduction

This diary is to help you keep a personal record of what is happening to you.

In its pages you will find information to help you and your family.

It will not give you all the answers, but it will help you to:

- Find the information you need
- Know how to get the help and support that is right for you
- If you need help speaking with your family, there is a letter at the end of this booklet

You can use the booklet to keep a record of your results, your treatment and what is planned next.

You can ask any member of the professional team looking after you to write in it for you, or you can write the information yourself.

Fabry

RECORD BOOKLET

- Please bring this booklet to all hospital appointments and any admissions to A&E
- Show the booklet to your GP, dentist, pharmacist, home therapy nurse or Fabry specialist nurse

- NAME .....
- HOSPITAL .....
- HOSPITAL NUMBER .....
- CONSULTANT .....
- HOMECARE TEAM .....

Appointments

Date	Time	Place, purpose and likely duration

Appointments

Date	Time	Place, purpose and likely duration

Contents

Key

- ■ caption to fill in
- **T** phone number to fill in
- **B** bleep number to fill in

Contact us if you have a problem associated with your treatment or condition

If you notice any of these effects during or after your treatment you should tell your doctor immediately:

Reaction to ERT
Chills, fever, feeling cold, nausea, vomiting, headache, abnormal feelings in the skin such as burning or tingling, sweating, fast heart rate, light-headedness, hives, swelling in your hands, feet, ankles, face, lips, mouth or throat which may cause difficulty in swallowing or breathing.

Reaction to chaperone therapy
Headache.

Refer to page 46 for a full list of very common and common potential side effects associated with ERT and chaperone therapy.

Further information on your treatment is available in the information provided by your healthcare team.

If you experience any of the above reactions or feel unwell during an enzyme infusion or whilst taking chaperone therapy

1. Stop the infusion (if applicable)
2. Follow your personal reaction protocol
3. If symptoms persist, contact the Lysosomal Storage Disorders Unit (LSDU) or your home care nurse.If symptoms are severe/very severe or if it is a medical emergency, call 999

Reporting of side effects
If you get any side effects, talk to your specialist healthcare team. This includes any possible side effects not listed in the package leaflet. You can also report side effects directly via the Yellow Card Scheme at: www.mhra.gov.uk/yellowcard or search for MHRA Yellow Card in the Google Play or Apple App Store.
By reporting side effects you can help provide more information on the safety of this medicine.

Useful telephone numbers

Useful telephone numbers and bleep numbers

On-call doctor (out of hours)	T	.....
	B	.....
Nurse specialist	T	.....
	B	.....
Secretary	T	.....
	B	.....
Other	T	.....
	B	.....

Community team telephone numbers

General practitioner	T	.....
District nurse	T	.....
Home care nurse	T	.....
Other	T	.....

Your treatment details and information

Diagnosis:

- TREATMENT PROTOCOL
  - ERT/CHAPERONE THERAPY:
  - OTHER:
  - DOSE:
  - FREQUENCY:
- PREMEDICATIONS
  - DRUG:
  - DOSE:
  - FREQUENCY:
- CLINICAL TRIAL CODE (if applicable)
- PROPOSED DURATION OF TREATMENTS
- DATE TREATMENT STARTED
- DRUG SENSITIVITIES
- CONTRAINDICATED DRUGS DURING TREATMENT

Your treatment details and information for ERT if applicable

- VENOUS ACCESS DEVICE
- DATE DEVICE INSERTED
- WHAT TO DO IF YOU FEEL UNWELL PRIOR TO YOUR INFUSION

The effects of Fabry disease

Fabry disease is an inherited disorder that is one of a group called lysosomal storage disorders. Fabry disease results from abnormally low levels of an enzyme, alpha galactosidase A enzyme (α-GAL A), which normally breaks down a fat called globotriaosylceramide (Gb3) so that it can be recycled. When the enzyme is absent, the waste product (Gb3) builds up in various cells and organs in the body. A wide variety of signs and symptoms are associated with Fabry disease. A person with Fabry disease may not experience them all and the severity of symptoms can vary from person to person. However, as Gb3 builds up, health problems and symptoms tend to get progressively worse.

Pain

One of the first symptoms, which can occur even in childhood, may be a painful burning sensation in the hands and feet called acroparesthesia. The pain can be severe and worsen with exercise, stress, illness and variations in temperature.

Stomach and intestines

Early symptoms of Fabry disease can include abdominal cramps, frequent bowel movements shortly after eating, diarrhoea and nausea.

Skin conditions

Some patients (especially men and boys) have a red, non-painful rash, known as angiokeratoma, which appears usually in the area between the belly button and the knees. It can also appear on other parts of the body, such as the lips, tongue, hands and toes. It may be confined to a small area of the body, or affect a larger area.

Individuals with Fabry disease can also experience a decreased ability, or complete inability to sweat. This can lead to sensitivity to heat and difficulty coping with physical exercise. In contrast, some individuals may sweat too much.

Eyes

The surface layer of the eye (cornea) may appear abnormal when examined using special equipment that an optician or an eye specialist (ophthalmologist) may have. This faint circular pattern, which is called cornea verticillata, does not affect vision but may increase with time.

Ears

Fabry disease can affect hearing and patients may experience a ringing in the ears called tinnitus. Most patients will experience some degree of hearing loss, which can either progress over time or be quite sudden.

Kidney

The progressive build up of Gb3 can lead to kidney problems. This can lead to an abnormal amount of protein in the urine (proteinuria). Kidney problems occur early in life in a lot of children, in many women and in almost all men with Fabry disease. Severe kidney problems can occasionally lead to kidney failure, requiring dialysis or a transplant.

Heart

Heart abnormalities can include changes in the size of the heart (left ventricular enlargement), irregular heartbeat and leaky heart valves. Such problems can increase the risk of further heart complications.

Central Nervous System (CNS)

The progressive build up of Gb3 can lead to CNS symptoms including headache, vertigo/dizziness, vascular dementia, transient ischaemic attacks and stroke.

Enzyme replacement therapy (ERT)

What is ERT?

Enyzme replacement therapy (ERT).

ERT is indicated for long-term treatment in patients with a confirmed diagnosis of Fabry disease. However some ERTs are either not licensed or have not established safety and efficacy in younger children (under 8) or adults >65 years. Your doctor will be able to advise on whether a particular ERT is suitable for you.

ERT for Fabry disease is given as an intravenous infusion (where the medicine is administered into the vein slowly over a period of time, rather than as an injection). These infusions are given every 14 days. The treatments will be administered by a nurse, initially in hospital. Long-term management options will be discussed with your healthcare team, including home infusion and, if appropriate, the option for self-infusion.

The aim of ERT is to address the underlying enzyme deficiency in order to treat and prevent the complications associated with the disease.

The principle of ERT is to replace the deficient α-GAL A enzyme and, therefore, help prevent further accumulation of Gb3.

Two ERT treatments are currently available in the UK. Both are given by intravenous infusion every other week. Your doctor will discuss the options with you.

Very common side effects of ERT and what to do if you have a reaction

Like all medicines, ERT may cause side effects, although not everybody gets them.

If you notice any of these effects during or after an infusion, you should tell your doctor immediately:

Chills, fever, feeling cold, nausea, vomiting, headache, abnormal feelings in the skin such as burning or tingling, sweating, fast heart rate, light-headedness, hives, swelling in your hands, feet, ankles, face, lips, mouth or throat which may cause difficulty in swallowing or breathing.

Refer to page 46 for a full list of very common and common potential side effects associated with ERT.

Further information on your treatment is available in the information provided by your healthcare team.

Your doctor/nurse may stop the infusion temporarily (5–10 min) until the symptoms go away and then begin the infusion again.

Your doctor/nurse may also treat the symptoms with other medicines (antihistamines or corticosteroids).

Most of the time you can still be given your ERT, even if these symptoms occur.

If you experience a severe allergic (anaphylactic-type) reaction, the administration of your ERT will immediately discontinued and an appropriate treatment will be initiated by your doctor.

Chaperone therapy

What is chaperone therapy?

It is an oral medication known as a pharmacological chaperone and is indicated for the treatment of adults (and adolescents aged 16 years and older) with a confirmed diagnosis of Fabry disease with an amenable gene mutation. Amenable means a mutation where the medication has been shown to have a positive effect on enzyme activity.

The recommended dose is 1 capsule orally once every other day, at the same time of day.

It should be taken on an empty stomach with no food for at least 2 hours before and 2 hours after taking. Clear liquids, including carbonated drinks, can be consumed during this period.

Capsules should be swallowed whole and not crushed or chewed.

If a dose is missed entirely for the day, take the missed dose only if it is within 12 hours of the normal time that the dose should have been taken. If more than 12 hours have passed, resume taking it at the next planned dosing day and time and according to the every-other-day dosing schedule.

You should discuss with you doctor and nurse if you have a suitable mutation and would like to have chaperone therapy.

Further information on your treatment is available in the information provided by your healthcare team.

Very common side effects of chaperone therapy and what to do if you have a reaction

Possible side effects

Like ERT, chaperone therapy may cause side effects, although not everyone gets them.

Possible very common side effects include:

- Headache.

Refer to page 46 for a full list of very common and common potential side effects associated with chaperone therapy.

Further information on your treatment, including common side effects, is available in the information provided by your healthcare team.

It is important to discuss any side effects with you doctor/nurse at your specialist centre before stopping medication. You should also inform them if you miss any doses.

If your Fabry disease does not respond to treatment or you have ongoing side effects, this will be discussed with your specialist doctor and your treatment will be reviewed.

Reporting of side effects

If you get any side effects, talk to your specialist healthcare team. This includes any possible side effects not listed in the package leaflet. You can also report side effects directly via the Yellow Card Scheme at: www.mhra.gov.uk/yellowcard or search for MHRA Yellow Card in the Google Play or Apple App Store.
By reporting side effects you can help provide more information on the safety of this medicine.

This will be discussed with your consultant.

Treatment centre & charter for patients receiving ERT/chaperone therapy

Aim

This charter is intended to set out the ways in which the Lysosomal Storage Disorders Unit (LSDU) and its patients can work together to ensure that the LSDU is able to provide an efficient ERT/chaperone therapy service. It is intended to identify ways in which both parties can act in order to help each other.

It is our aim to provide the following services

- We will undertake to administer your ERT infusions or prescribe chaperone therapy on a regular basis in line with your treatment schedule
- We will arrange for transfer to the homecare nursing service at an appropriate time in your treatment as applicable
- We will make regular lysosomal storage out-patient clinic appointments for you
- We will make regular assessment appointments for you, at times that are both convenient to you and within our scheduled clinic times
- We will tell you the results of the assessments, both at the clinic and follow these up in writing
- We will discuss with you your response to ERT/chaperone therapy and whether any changes to your treatment regime are necessary
- We will discuss with you, in a sensitive and timely fashion, any withdrawal of treatment if there is evidence of continued disease progression despite regular therapy

What we ask of you

- To attend regular infusions and appointments either at a treatment centre or at home with the homecare nursing service
- To inform the relevant person as soon as possible if you can't attend an infusion or appointment
- To ensure that you comply with the terms and conditions of the homecare nursing service
- To ensure that those involved in your care have your correct contact details
- To ensure that you attend planned LSDU outpatient clinic appointments
- To ensure that you attend all assessments that are booked for you. If you cannot attend an appointment you should re-book it directly with the relevant department as soon as possible

Ongoing concerns

In the event of being unable to maintain these goals, you will be:

- Reminded by telephone
- If problems persist, we will send you a letter, which will also be copied to your GP
- ERT infusions or chaperone therapy may have to be discontinued if problems persist

If you feel that you will be able to comply with the above, please fill in your details below and sign.

Name:	Name of Clinician:
.....	.....
Signature:	Signature of Clinician:
.....	.....
Date:	Date:
.....	.....

Initial assessment tests

An initial assessment test is a way for our doctors to find out your medical history and medication (please bring a list with you). The doctor may also wish to examine you. You may meet one or more of our specialist doctors who have a particular interest in different organ functions (e.g. kidneys, bowel) and how they relate to Fabry disease.

Blood tests

During the initial assessment test, you will have about 20 mL of blood taken, which will measure various aspects of your health, such as your blood count, kidney function, liver function, cholesterol and vitamin levels.

Measurements of kidney function – glomerular filtration rate

Most patients have a measurement taken called a nuclear medicine estimation of glomerular filtration rate. This involves injecting a small amount of tracer and then, four hours later, taking a small blood sample to see how well the kidneys have removed the tracer from your body. The tracer is completely harmless. You will leave the department to perform other tests during the four-hour interval.

24-hour urine

We will have sent you a bottle for a 24-hour urine collection, so that we can assess your kidney function and measure the amount of protein in the urine. This will come with instructions as to how to collect the urine.

MRI of the brain

Most patients have magnetic resonance imaging (MRI) taken of the brain to look for the effects of Fabry disease. The procedure involves going in an imaging machine for approximately 30 minutes to give us detailed pictures of your brain. People with pace-makers or other metal implants are not able to do this and we will arrange an alternative scan called a CT scan. Please let us know if this applies to you.

Investigations and monitoring

Audiology

We will carry out an audiology test, which is a simple hearing test that you may have had in childhood. It will assess your ability to hear sounds of different pitches.

Eye test

Just like when you go to the opticians, the eye doctor may request putting some drops in your eyes to obtain a better view.

Heart assessment

You will have an electrical measurement of the heart (ECG); this causes no discomfort and takes about five minutes. You will also have an echocardiogram, which involves using an ultrasound scan to look at your heart. You may also have an exercise test and an MRI scan of your heart if the cardiac specialist doctor asks for this.

You will be given more detailed information regarding all of these tests when you visit and we will assist you in finding the location and times of all of the appointments.

If you have any questions before your visit, please do contact us.

My treatment goals and ERT/chaperone therapy starting criteria

My treatment goals:

1. .....

2. .....

3. .....

Starting criteria for adults

In males with "classical mutations" (leucocyte enzyme activity <5% and classical phenotype) Fabry-specific therapy should be considered at diagnosis. In adult females and males with 'later onset', Fabry-specific therapy should commence when one of the following criteria are fulfilled:

1. **Evidence of Fabry related renal disease (one of):**
   - Chronic kidney disease (CKD) stages 3: at least 2 consistent estimates or measured GFR over a minimum of 6 months ☐
   - CKD stage 2: at least 3 consistent estimates or measured GFR over at least 12 months with a GFR slope greater than age-related normal ☐
   - Persistent Proteinuria >300 mgs/24 hours for males. Females seldom progress to ESRF. In female, if proteinuria is the only presentation – anti- proteinuria medications (ACE/ARB) should be tried in the first instance for a minimum period of 12 months ☐

2. **Evidence of Fabry related cardiac disease (one of):**
   - LV wall thickness >13 mm in males and >12 mm in females ☐
   - LV mass index by 2D echo/CMR above normal for age and sex ☐
   - Late gadolinium enhancement on cMRI ☐

My ERT/chaperone therapy starting criteria

3. General symptoms of Anderson-Fabry disease

- Uncontrolled pain or gastrointestinal symptoms leading to a need to alter lifestyle or interferes with quality of life ☐
- Patients whose sole eligibility criterion is pain should have been assessed by a specialist pain team. Patients whose sole eligibility criterion is GI symptoms should have been assessed by a specialist GI team ☐
- If Fabry related symptoms is the only indication for consideration of Fabry-specific therapy a trial could be given for a year with a pre specified outcomes agreed by the treating physician and the patient as to what would constitute a positive effect for symptom control. Such outcomes may include:
  - Reduction in the need for analgesics ☐
  - Reduction in time lost from work ☐
  - Significant improvements in validated pain scoring and or quality of life measures ☐

Baseline assessment

Date Regimen

Results

Symptoms

GFR

Urine protein

Echo

MRI brain

Audiogram

Vitamin D

Ferritin

Other

Oral medications

Special instructions/comments

Test or scan	Date	Time	Place of test	Form
Tests/scans needed before next treatment				

Follow Up 1

Date Regimen

Results

Symptoms

GFR

Urine protein

Echo

MRI brain

Audiogram

Vitamin D

Ferritin

Other

Oral medications

Special instructions/comments

Test or scan	Date	Time	Place of test	Form
Tests/scans needed before next treatment				

Follow Up 2

Date Regimen

Results

Symptoms

GFR

Urine protein

Echo

MRI brain

Audiogram

Vitamin D

Ferritin

Other

Oral medications

Special instructions/comments

Test or scan	Date	Time	Place of test	Form
Tests/scans needed before next treatment				

Admissions

Date admitted	Date discharged	Reason for admission

Other out-patient appointments

Date	Hospital	Clinic	Outcome

Other medication

These next pages are for you to make notes on any medication you are currently taking, why you are taking them and how often. You may also want to record any over-the-counter medicines or remedies you take.

Medication name	Date started	Date stopped	When to take it	What is it for?	Notes/comments

ERT infusion dates (if applicable)

Please record the date of each infusion

Date	Dose	Comments	Actual dose	Expected interval	Expected number of infusions prior to next visit

ERT infusion dates (if applicable)

Please record the date of each infusion

Date	Dose	Comments	Actual dose	Expected interval	Expected number of infusions prior to next visit

ERT infusion dates (if applicable)

Please record the date of each infusion

Date	Dose	Comments	Actual dose	Expected interval	Expected number of infusions prior to next visit

ERT infusion dates (if applicable)

Please record the date of each infusion

Date	Dose	Comments	Actual dose	Expected interval	Expected number of infusions prior to next visit

Information for relatives

This page includes an example letter that can be sent to inform your family members of your diagnosis with Fabry disease, including advice on next steps they can take.

Dear family member/s,

I am writing to let you know that several/one of your/our family members have been diagnosed with Fabry disease. Fabry disease is a metabolic condition that results from a deficiency of the alpha-galactosidase A (alpha-GAL) enzyme. This enzyme is necessary to break down certain fatty substances for reuse by the cell or elimination from the body. Without enough alpha-GAL enzyme, these fatty substances (primarily Gb-3) progressively accumulate in cells throughout the body. Fabry disease is an X-linked recessive disease, thus men are more often affected with 'classic Fabry disease', however, female "carriers" of the disease may also be affected.

Fabry disease affects multiple organ systems; affected individuals typically experience burning or pain in their hands and feet at some time in their lives. Gastrointestinal problems, such as pain and bloating, decreased appetite or diarrhoea, are common. The ability to sweat may be decreased or altogether absent, making exercise or exposure to heat intolerable. Angiokeratomas, small reddish purple lesions, may be found on the skin. A circular pattern on the surface layer of the eye (cornea) may be noted on a slit-lamp eye exam. Decreased energy levels, frequent fevers and headaches are also common symptoms. Patients with Fabry disease are at risk of renal failure, stroke and heart disease.

Please understand that each of these symptoms listed above may occur in patients not affected with Fabry disease, this letter is not offering a diagnosis or statement of risk. I simply wish to inform you, as a family member related to someone with Fabry disease, of your potential risk of inheriting this disorder. I would encourage you to get genetic counselling to discuss Fabry disease, its genetic inheritance pattern and to help you decide whether to be tested to determine your risk. Treatment options for Fabry disease are available for appropriate patients, so identifying those with Fabry is really important.

Testing to determine whether or not you are affected with Fabry disease is done on both a research and clinical basis. A simple blood sample can be used to perform enzyme and DNA analyses.

If you have questions or concerns about Fabry disease, or if you wish to arrange genetic counselling, diagnostic/medical evaluation and/or testing, please contact the treatment centre from which your family member is receiving treatment. Staff at the unit will be happy to provide you or your primary care physician with information about diagnostic testing, inheritance risk and treatment.

Patient association website

www.mpssociety.org.uk

The society for Mucopolysaccharide Disease (the MPS Society) is a UK charity supporting individuals and families affected by MPS, Fabry and related diseases.

General Fabry information

A

Acroparesthesia – Tingling sensations or numbness in the extremities (e.g. hands, fingers, toes).

Alpha-galactosidase A (α-GAL A) – An enzyme that helps to break down globotriaosylceramide (Gb3).

Analgesic – Any drug that relieves pain. Aspirin and paracetamol are mild analgesics.

Anaemia – A condition in which a decreased number of red blood cells may cause symptoms including tiredness, shortness of breath and weakness.

Anderson-Fabry disease – A lysosomal storage disorder (LSD) named after the doctors who first identified the disease; it is also known as Fabry disease.

Angiokeratoma – A skin rash, which appears as small, raised, dark-red spots.

Anhidrosis – An inability to sweat.

Antiemetic agent – A drug that prevents or controls nausea and vomiting.

B

Blood cells – Minute structures produced in the bone marrow; they consist of red blood cells, white blood cells and platelets.

Blood count – The number of red blood cells, white blood cells and platelets in a sample of blood.

C

Cardiac – Relating to the heart.

Central line or central venous catheter – A special intravenous tubing that is surgically inserted into a larger vein near the heart and exits from the chest or abdomen. The catheter allows medications, fluids or blood products to be given and blood samples to be taken.

(Examples of central venous catheters are Broviac, Groshong, Hickman, etc.).

Chaperone therapy – A treatment that uses small molecules to facilitate the proper formation of mutated proteins.

Cornea – The transparent front part of the eye that helps the eye to focus.

Cornea verticillata – A whorl-like opaque area on the cornea, which can be seen during an eye examination when using special equipment.

D

Dialysis – The process of removing waste products from the blood using a special machine.

E

Enzyme – A protein that helps to speed up chemical reactions in living organisms.

Enzyme replacement therapy (ERT) – A type of treatment that replaces a deficient enzyme in the body.

General Fabry information

F

Fabry disease – A lysosomal storage disorder (LSD); it is also known as Anderson-Fabry disease.

Full blood count – A full blood count (FBC), or complete blood count (CBC), is a test that gives information about the cells in a patient's blood. An FBC will normally include red cells, white cells and platelets.

G

Gastrointestinal – Relating to the digestive system (e.g. stomach, large and small bowel).

Globotriaosylceramide (Gb3, GL3 or CTH) – A fatty substance that is normally broken down by alpha-galactosidase A and builds up in the body in patients with Fabry disease.

H

Haemoglobin (Hb) – The substance inside red blood cells that binds to oxygen and carries it from the lungs to the tissues.

Haemorrhagic stroke – A type of stroke caused by a blood vessel bursting, which interrupts the flow of blood in the brain.

Hereditary – The transfer of characteristics from a parent to a child.

Hyperhidrosis – An abnormal increase in sweating.

Hypohidrosis – An abnormal decrease in sweating.

I

Infusion – The process of introducing a fluid into the body, usually through a vein (intravenous infusion).

Infusion pump – A device that delivers measured amounts of fluids or medications into the bloodstream over a period of time.

Injection – Pushing a medication into the body with the use of a syringe and needle.

Intramuscular (IM): Injection into a muscle.

Intravenous (IV): Injection into a vein.

Subcutaneous: Injection into the fatty tissue under the skin.

Irritable bowel syndrome – A chronic disorder of the digestive system, which causes pain and discomfort.

Ischaemic stroke – A type of stroke caused by a blockage that stops blood from reaching the brain.

L

Left ventricular enlargement – An abnormal increase in the size of one of the chambers of the heart.

Lysosomal storage disorders (LSDs) – A group of rare genetic medical disorders caused by defects in the lysosomes; also known as lysosomal storage diseases.

Lysosome – A subunit of human cells that contains enzymes to help break down waste products.

General Fabry information

M

Metabolic disease – A medical disorder that affects the production of energy in human cells.

Molecule – The small unit of a substance.

N

Nausea – Feeling sick.

Nephrologist – A doctor who specialises in problems of the kidneys.

Neurologist – A doctor who specialises in problems of the nervous system (including the brain, spinal cord and nerves).

O

Ophthalmologist – A doctor who specialises in problems of the eyes.

Opiate-based pain relief – A medicine that is given to reduce severe pain and is related to morphine.

P

Pacemaker – A medical device that helps to regulate the heartbeat.

Porta-cath – A catheter connected to a quarter-sized disc that is surgically placed just below the skin in the chest or abdomen. The tube is inserted into a large vein or artery, directly into the bloodstream. Fluids, drugs or blood products can be infused, and blood can be drawn through a needle that is stuck into the disc.

Protein – A large molecule that forms an essential part of all living cells; all enzymes are proteins.

Proteinuria – An abnormal increase in protein in the urine.

R

Renal – Relating to the kidneys.

S

Side effects – Secondary effects of drugs used for disease treatment.

Sign – A change caused by a disorder that is seen by a clinician but not always noticed by the patient.

Steroids – A type of hormone.

Stroke – A loss of brain function caused by the interruption of the blood supply to the brain.

Symptom – A change caused by a disorder that is noticed by the patient.

T

Tinnitus – A ringing sound in the ear.

V

Venipuncture – Puncturing a vein in order to obtain blood samples to start an intravenous drip or to give medication.

Vertigo – A sensation of dizziness or disorder of balance.

This is for you to show your GP or staff at A&E

This patient has been undergoing the following treatment

☐ ERT treatment ☐ chaperone therapy

for Fabry disease

at: .....

Please do not hesitate to contact the specialist centre if you have any concerns.

- Palpitations
- Chest pains
- Symptoms suggestive of weakness, numbness, severe headache, suggestions of transient ischaemic attack or stroke

Please do not hesitate to contact us for advice. Ring the hospital switchboard and ask for the registrar – see the front of this booklet to identify the appropriate consultant team. During office hours, if the registrar is not available, ask for the specialist nurse.

Potential treatment side effects

As with any medicine, you may experience side effects with ERT or chaperone therapy - these are listed below.

ERT

Severe life-threatening allergic reactions ("anaphylactoid reactions") have been reported in some patients. If you experience any serious side effect, you should contact your doctor immediately.

There are two ERTs available. The following list combines the possible side effects for these two ERTs. Please speak to your healthcare professional for the full list of possible side effects specific for each treatment.

Very common

Swelling in the tissue (e.g. legs, arm), dizziness, tingling or numbness or pain in fingers or toes, tremor, ear ringing, palpitations (the feeling of a pounding heart), shortness of breath, cough, sore throat, abdominal pain, diarrhoea, rash, back or limb pain, muscle pain, joint pain, chest pain, cold symptoms, fever, chills, feeling cold, feeling tired or weak, feeling sick (nausea), vomiting, headache and abnormal feelings in the skin such as burning or tingling.

Common

Change in the taste of food, prolonged sleep, eyes tearing, increased ear ringing, increased heart rate, heart rhythm problems, increased blood pressure, low blood pressure, facial flushing (redness), hoarseness, or tight throat, runny nose, abdominal discomfort, acne, red or itchy or mottled skin, excessive sweating, muscle and bone discomfort, swelling of the extremities or joints, hypersensitivity, chest tightness, increased feeling of lack of energy, feeling cold or hot, flu-like symptoms, discomfort, chest pain, difficulty breathing and exacerbated difficulty in breathing, pale appearance (pallor), itching, feeling weak, nasal congestion, diarrhoea, muscle pain, sudden swelling of face or throat, oedema (swelling) in extremities, sensation of spinning (vertigo), muscle spasms, sleepiness, abdominal pain, back pain, rash, low heart rate, fainting, cough, joint pain, face oedema, muscle tightness, pain, dizziness, palpitations, decreased sensitivity to pain, burning sensation, wheezing, pain at the extremities, nasopharyngitis, hyperthermia, decreased mouth sensitivity, musculoskeletal stiffness.

Chaperone therapy

Very common

Headache.

Common

Palpitations (the feeling of a pounding heart), sensation of spinning (vertigo), diarrhoea, feeling sick (nausea), stomach ache, constipation, dry mouth, sudden need to defecate, indigestion (dyspepsia), tiredness, raised levels of creatine phosphokinase in blood tests, weight gain, muscle spasms, muscle pain (myalgia), pain in the extremities, painful stiff neck (torticollis), tingling in extremities (paraesthesia), dizziness, reduced sense of touch or sensation (hypoaesthesia), depression, protein in the urine (proteinuria), shortness of breath (dyspnoea), nose bleed (epistaxis), rash, persistent itch (pruritus) and pain.

Useful organisations

Addenbrooke's Hospital
Cambridge
Tel: 01223 274 634

Belfast City Hospital
Belfast
Tel: 028 9026 3874

Birmingham Children's Hospital
Birmingham
Tel: 0121 333 9999

Great Ormond Street Hospital
London
Tel: 020 7405 9200

Manchester Children's Hospital
Manchester
Tel: 0161 701 2137/8

National Hospital for Neurology and Neurosurgery
London
Tel: 020 3448 4778

Queen Elizabeth Hospital
Birmingham
Tel: 0121 627 2000

Royal Hospital for Sick Children
Glasgow
Tel: 0141 201 0000

Salford Royal NHS Foundation Trust
Manchester
Tel: 0161 206 4365

The Royal Free Hospital
London
Tel: 020 7472 6409

The Royal Hospital for Sick Children
Edinburgh
Tel: 0131 536 0000

University of Wales Hospital
Cardiff
Tel: 029 2074 3275

Patient organisation

MPS Society
MPS House, Repton Place
White Lion Road
Amersham
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