NHS
Birmingham Women's and Children's
NHS Foundation Trust

Wolfram

Information Book

Wolfram Information Book

Contents

Wolfram Syndrome Information Book This information has been written so that you can keep a brief record of your child’s condition and care.

It is meant to be used to share information between yourselves and all health care professionals, both in hospital and the community. This is a record which you can add to when you see or receive relevant information.

This pack is your property, and your responsibility, therefore you may choose who sees the information recorded.

Take it to every out-patient appointment, hospital visit, school, medical, dental visit and health visitor check. Also take it on holiday with you as a source of information in emergency cases.

Personal Details

Name:

I liked to be called:

PID: Date of Birth: Sex:

Address:

Telephone number:

Mobile:

E-Mail address:

Parent/Care:

Sibling names:

Language:

Interpreter required:

Religion:

School:

Telephone No:

School address:

Teacher:

Further Details

GP Information

GP Name:

Telephone No:

GP Address:

BWC Consultants

Wolfram consultant:

Wolfram Nurse:

Local Consultants

Endocrinologist:

Diabetes specialist nurse:

Ophthalmologist:

Audiologist:

Urologist:

Neurologist:

Others:

How to contact us:

The Wolfram team are available 09:00hrs to 17:00hrs Monday to Friday by contacting the hospital on:

0121 333 9272

Ask for:

• Wolfram Syndrome Co-Ordinator

• Wolfram Consultant

• Wolfram Nurse

In an emergency at any time call:

0121 333 9999 and ask for Endocrine Consultant on call

Alternatively, you can email us on diabetes.results@bch.nhs.uk.

The Wolfram Family Support Coordinator also supports families in their local area, working with professionals and organisations to make sure the families are getting the help they need, and can be contacted on.

Email: georginaking@wolframsyndrome.co.uk

Phone: 07592 629813

Adult team at University Hospital Birmingham:

Wolfram (WS) Secretary 0121 371 6845

Wolfram (WS) Consultant 0121 627 2000 (hospital switchboard)

WS Adult Support Co-ordinator

Email: phillippafarrant@wolframsyndrome.co.uk

Mobile: 07752 193635

What is Wolfram Syndrome?

Background

Wolfram Syndrome (WS) is a rare genetic disorder, which is sometimes referred to as DIDMOAD after its four most common presentations:

- Diabetes Insipidus
- Diabetes Mellitus
- Optic Atrophy
- Deafness.

Individuals with WS can also experience problems with the urinary tract, neurological symptoms and sometimes there are problems with the digestive system.

Every person affected is different and may only have two or three of the main features.

What are the symptoms?

WS is characterised by:

Diabetes Insipidus – This is where the body cannot concentrate urine because the posterior pituitary gland (found at the base of the brain) isn't making enough of the hormone vasopressin. This means you get very thirsty and need to pass urine frequently.

Diabetes Mellitus – is the name given when the body cannot convert glucose into energy, because the pancreas is not making enough of the hormone insulin. Symptoms include thirst, frequent

passing of urine and weight loss. Insulin injections are essential to treat this form of diabetes.

Optic atrophy (nerve damage to the eye). Symptoms often present as colour blindness, difficulty seeing in the classroom at school, or everything going grey.

Deafness – It can be difficult to hear in a crowded room, or to hear high pitched sounds.

Renal problems may occur including urinary tract disorders and bladder problems, this can cause bedwetting, needing to pass urine frequently and loss of bladder control. This is known as a neurogenic bladder.

Neurological problems - These can include loss of balance, sudden muscle jerks, loss of taste and smell, breathing problems and depression. Choking/ swallowing problems may also occur.

Other features of WS may include fertility problems and gastrointestinal problems causing problems with swallowing, constipation or diarrhoea. Not all of the symptoms listed above are necessarily present in those diagnosed with WS, and each one can vary in severity and onset.

What are the causes?

It is an inherited genetic condition. A change in the WSF1 gene (2 misprints are found, a bit like a spelling mistake) causes more than 90% of WS.

We are now starting to see some individuals with a few of the problems we expect to see in WS, but the pattern of these features is a bit different. Some of these individuals may have a ‘misprint’ in one copy of their WFS1 gene and we say they have Wolfram syndrome-like disease.

The WSF1 gene provides instructions for producing a protein called Wolframin. This protein is used within all cells of the body.

The change in WSF1 gene leads to the production of a Wolframin protein that has reduced or absent function.

When the cells do not have enough functioning Wolframin, the cells trigger their own ‘cell death’.

What are the chances of having another affected child?

WS is inherited as an autosomal recessive condition; this means that both parents carry one abnormal copy of the Wolfram gene, and one normal copy. For a child to be affected, he/she has to inherit two abnormal copies, one from each parent. The chances of parents having another affected child are about 25%.

It is possible to test if an unborn child is affected during pregnancy.

How do you tell if you have this condition?

If insulin-dependent diabetes mellitus and optic atrophy symptoms are present by fifteen years of age, WS is presumed to exist. Genetic testing can confirm the presence of WS.

How is it treated?

The condition is managed by treating the symptoms as they appear. Hormone therapy, such as vasopressin in a nasal spray, or tablets may relieve the symptoms of thirst and frequency in passing urine caused by diabetes Insipidus. Management of diabetes mellitus can be achieved by insulin injections/or insulin pump, blood glucose monitoring and healthy eating. There is no known treatment for optic atrophy, but visual aids such as magnifiers and speech software may help. A hearing aid may be used to help with hearing loss, in some cases cochlear implants may be a possibility. Psychotherapy and antidepressants may be used to treat depression. Some of the neurological symptoms can be treated with medication. Renal problems may be treated by catheterization (passing a thin flexible tube into the bladder to drain away urine), or tablets.

Will there be a cure for WS?

Any cure is a long way off. The current research is to understand why mistakes in the WSF1 gene cause the syndrome. There are research groups in America, UK, France, Italy, Belgium, Germany and Japan all investigating this problem.

Where can I go for help?

Wolfram Syndrome UK supports families and individuals living with WS www.wolframsyndrome.co.uk. There is a WS website where you can register and contact other families affected all over the world. www.wolframsyndrome.org

The WSUK family support coordinator supports families attending clinics and in other ways outside the clinics.

There is a global patient registry that is free to sign up to https://wsglobalregistry.iamrare.org/.

WOLFRAM SYNDROME (WS)

A very rare genetic condition that affects 1 in 770,00 of the UK population

There are around 120 people in the UK with WS

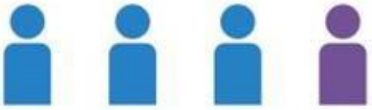
It affects 1:550,000 children In the UK

Parents of a child with WS have one abnormal copy of WSF1 and one normal copy

Both parents must pass on one abnormal copy of WSF1 for a child to be affected

It is caused by a mistake in the WSF1 gene

There is a one in four chance of a sibling having the condition



WS is so rare many doctors will not be aware of it

Support is available through Wolfram Syndrome UK

WOLFRAM SYNDROME UK
Inform, Support, CURE

Symptoms can include:
Diabetes
Vision loss
Hearing loss
Renal problems
Neurological problems

Not everybody will get these symptoms. Some can be managed with medication.

There is no cure at present, however ongoing research means there may be clinical trials into a therapy in the very near future.

What is Diabetes Mellitus?

Diabetes Mellitus occurs when the cells in the pancreas (an organ in your body) that normally produces insulin are damaged. The pancreas produces less insulin than normal or no insulin at all.

When we eat food containing carbohydrate (starch and sugar) it produces glucose, which is transferred from the stomach into the blood stream and then into the cells to provide energy and maintain normal blood glucose levels.

Insulin is responsible for this process and acts like a key. It opens the door to the cells in your body and allows the glucose to enter. The body's cells then convert the glucose into energy. Without insulin, more and more glucose will build up in the blood stream and your body will try to remove the glucose. This can lead to a chain of events including the following:

• The body will try to get rid of the excess glucose by pushing it out in the urine– resulting in the need to go to the toilet more often • Because more urine is been passed, there is increased thirst • Because your cells are not getting the energy they need, you will feel tired • Because your body still needs energy, it will start to break down fat stores, so there will be weight loss • If this continues for any length of time, there will be a build-up of ketones, which are toxic to the body. This condition is called Diabetic Ketoacidosis or DKA.

Treating Diabetes Mellitus

To maintain 'normal' blood glucose levels you need to do the job of your pancreas. This means injecting insulin several times a day, or if on insulin pump therapy, giving regular boluses - regularly monitoring your blood glucose levels and making constant decisions about how much insulin to inject to keep your blood glucose as close to normal range as possible. One of the reasons to do this is to help you feel your best and give you more energy.

Blood glucose monitoring

This is the best method of being in control of your diabetes, as it enables you to see how well your body is responding to your insulin, diet and activities, which in turn helps to keep you healthy and safe.

By providing “up to the minute” information on your blood glucose levels you can check that they are within range, or if you are at risk of a ‘hypo’ (low blood glucose) or ‘hyper’ (high blood glucose). You can then make immediate or day-to-day adjustments to your diabetes management.

Monitoring your diabetes

• Regular blood glucose monitoring and documenting your results in a diary will allow you to identify patterns of high or low blood glucose levels, which will help your diabetes team make the right insulin adjustments

• You should aim to do 4-6 blood glucose checks per day, this should include monitoring blood glucose levels before meals, 2 hours after meals, overnight and before, during and after exercise.

• Checking for blood ketones if your level is 14mmol/L or higher and at times of illness. You can do this by using a blood glucose meter that also measures ketones.

Improving blood glucose control

• Keep a good record of your blood glucose levels

• Review on a regular basis to look for patterns of highs/lows

• Contact your diabetes team to discuss any concerns

Blood glucose targets

The aim is to achieve blood glucose levels between 4-7mmol/L, however check with your diabetes team for your individual blood glucose targets.

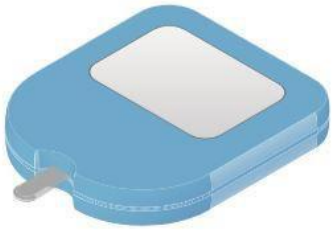
Using a blood glucose meter

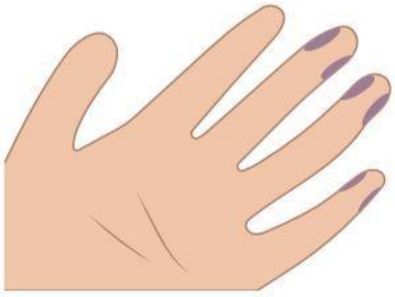
There are many blood glucose meters available and some of these have a talking function which can be particularly helpful if you are struggling to see the numbers on your meter screen. See your diabetes nurse for more information.

Good testing technique

It is important that blood glucose tests are carried out properly as poor technique may lead to incorrect results and inaccurate insulin dosing.

- Wash hands thoroughly with warm soap and water and dry thoroughly





- Take out a test strip and insert into the meter.

• Using the lancing device obtain a blood sample from the side of the finger, avoid thumb and index finger.

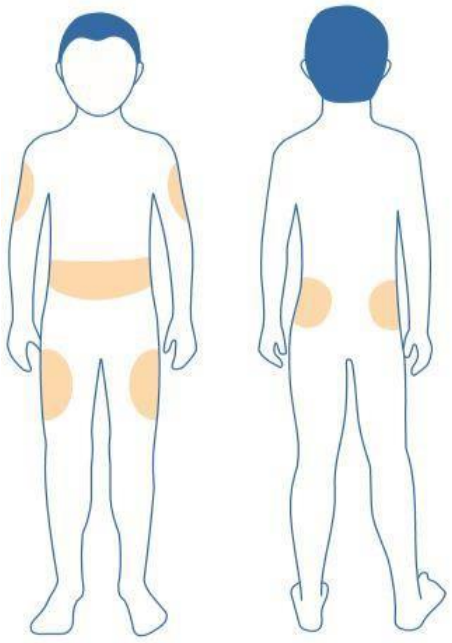
• Ensure to change the needle after each use, and vary the finger from which blood is taken.

• If you are struggling to get enough blood, turn finger upside down and squeeze the end of your finger (as though milking it), and wipe the first drop of blood away, squeeze again and place the blood onto the test strip and wait for result.

• Dispose of the test strip in the clinical waste bin and record the result in your diary.

Insulin Injections

Insulin injections are given into the fat just under your skin. The abdomen (tummy) is the most common site for injecting insulin; however other areas can be used, including the outside of your thighs and upper buttocks.



If the same place is used for each injection the area will become lumpy. These lumps will stop your insulin working properly, which will affect your blood glucose control.

If lumpy areas form they can take at least 6-8 weeks to go away, so it is a good idea to check for "lumpy" injection sites regularly.

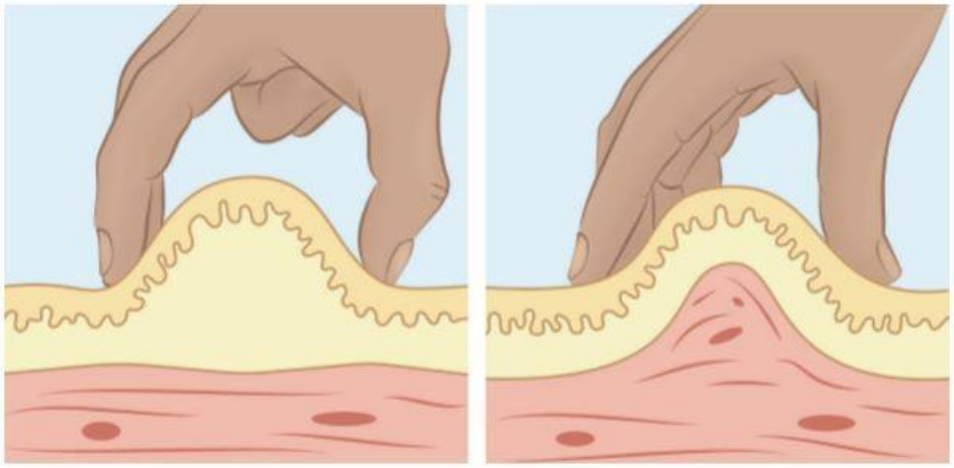
How to give the injection

• Attach a new needle each time.

• Hold the pen with the needle pointing upwards, dial up the units, then press the plunger to shoot some insulin into the air to make sure it works. This is called priming the pen.

• Dial up required amount of insulin units.

• Lift a skin fold (see picture), insert needle into selected injection site at a 90-degree angle.



• When inserted, push plunger fully until the dial reaches zero and hold for 10 seconds.

• Release skin fold.

• Remove pen.

• Remove needle and dispose of in sharps bin.

• Needles should only be used once

• If insulin leaks out of the injection site, you may not have injected deeply enough. If this happens often you may need to hold the needle in for longer than 10 seconds.

Aids available to help with injections

There are a variety of pens available that have clicking devices (if you find it difficult to see the numbers on the dial) so that you can count the clicks to know how many units you are injecting.

There are pre-filled disposable pens available for some brands of insulin, this means you do not have to re-load the pen when the insulin runs out.

Magnifiers are available to fit onto some insulin pens.

Insulin pumps

If you are struggling to input your carbohydrates into your pump then you can choose to safely deliver a bolus without looking at the screen by using the additional audio bolus feature.

Advice and setting up of this feature can be discussed with your diabetes team.

Hypoglycaemia

Hypoglycaemia (or hypo) is a low blood glucose level (below 4mmol/l). When this happens your body does not have enough energy to carry out its activities.

Why does hypoglycaemia happen?

- Too much insulin
- A delayed or missed meal or snack
- Sudden or sustained exercise
- Illness
- Anxiety
- Cold weather

• Injection sites

• Timing of insulin

The brain needs glucose as its source of energy. If the brain cannot store glucose it depends on a continuous supply from the blood, lowering of glucose levels can quickly interfere with brain function.

What are the symptoms (signs) of hypoglycaemia?

It is important that you check your blood glucose level to confirm the hypo.

Hypos can be grouped into 3 levels: mild, moderate and severe

Mild Hypo

A mild hypo is where you can treat yourself, and there are usually early warning signs. These hypos can be treated by taking some glucose by mouth as soon as the signs are noticed. Your diabetes nurse will advise you on the appropriate treatment.

Moderate Hypo

This is where you require help and is usually indicated by behaviour changes e.g. moodiness, aggression, and tearfulness. At this stage it is quicker to give glucose in a liquid form.

Glucogel is the best treatment at this stage.

Severe Hypo

A severe hypo is classed as loss of consciousness, being unresponsive or fitting.

Do not attempt to give anything by mouth. The best treatment is a Glucagon injection. This should be stored in the fridge at home.

How can I prevent a Hypo?

• Check your blood glucose regularly so you will know if you are going low.

• Know what the symptoms of a hypo are so that you can recognise and treat it early.

• Eat regularly and don’t miss meals.

• Eat some carbohydrates before and after you do exercise. (Discuss any exercise with your diabetes nurse as they will offer advice on insulin adjustments).

• If you are ill and unable to eat you are more likely to have a hypo, so will need to check your blood glucose more often and should have sugary drinks to prevent your blood glucose from dropping. (Speak to your diabetes nurse as they may advise a reduction in your insulin).

• Be prepared

• Always have dextrose tablets, Lucozade and glucogel at home and school

• Always carry dextrose tablets/ drinks with you

• Check your glucagon injection is in date

• Make sure your friends know you have diabetes and your hypo signs

• Always carry identification saying you have diabetes so that other people may help you.

Hyperglycaemia

Hyperglycaemia (high blood glucose) occurs when blood glucose levels get too high.

Reasons for hypoglycaemia:

• Not enough insulin

• Too much food/or incorrect carbohydrate counting

• Illness

• Stress

• Growth

• Reduced exercise

• Insulin injected at the wrong time

• Lumpy injection sites

Signs and symptoms of high blood glucose include:

Or you may have no signs at all.

Diabetic Ketoacidosis

What are ketones?

When there is not enough insulin in the body, or when the body is under stress such as during an infection or illness, glucose in the blood cannot enter the cells to provide energy and the body starts to break down body fat for energy. This leads to ketone production.

If the blood glucose levels rise and the body produce too many ketones and they make the blood too acidic. This is known as diabetic ketoacidosis, which is very dangerous and may require hospital admission.

Illness and diabetic ketoacidosis

Illness and infections can make the body more resistant to insulin, so you will require additional insulin. In addition, illness can cause

stress hormones to be released and these can cause blood glucose levels to rise.

Ketoacidosis can take a few hours to several days to develop. Recognising and treating it in the early stages is essential.

Warning signs of Ketoacidosis:

- Increased thirst
- Passing more urine
- Abdominal pain
- Drowsiness
- High blood glucose levels
- Ketones in your blood are over 1.5mmol/L
- Nausea or vomiting
- Rapid breathing
- Sweet smelling breath
- Confusion

Testing for ketones

If your blood glucose levels are over 14 mmol/L or you are feeling unwell, then you should check your ketones using your blood glucose meter. Contact your diabetes team if your blood ketones are over 0.6mmol/L.

Delayed treatment for high blood ketones level can be life threatening. Identifying rising ketone levels early can prevent an emergency situation or hospital admission.

Sick Day Rules

- **NEVER STOP INSULIN INJECTIONS or the INSULIN PUMP**

- The insulin dose may need increasing or decreasing, according to the results of blood glucose and ketone tests. For most illnesses more insulin is needed.

- Correction doses should be given until blood glucose is within normal range and ketones are less than 0.6mmol/L.

- Check blood glucose at least 3-4 hourly including through the night.

- If blood glucose is out of the normal daily target ranges or ketones are present you will need to check blood glucose 1-2 hourly, including through the night.

- Check blood ketones at the onset of any illness and then continue to check regularly if BG high (≥14 mmol/L) or vomiting (2 hourly).

- Take plenty of rest. Do not exercise as this can cause Diabetic ketoacidosis by increasing the body's demand for insulin at a time when the body is trying to cope with an illness

- Replace food with small amounts of carbohydrate containing liquids if not hungry

- Drink plenty of fluids to avoid dehydration. Taking fluids slowly and steadily reduces the likelihood of vomiting. If vomiting is only intermittent some of the fluids and carbohydrate taken in is still likely to have been absorbed. This can be checked by monitoring the blood glucose and ketones levels.

- Contact your diabetes team immediately if you are vomiting or unsure how to manage your high blood glucose and ketones.

What service should you expect?

We aim to offer support for children who are diagnosed with or suspected to have WS or related conditions. We do not replace care by your local hospital and community teams, but we do try to be a source of expert advice offering medical and emotional support for children and their families. The purpose of this service is to improve the quality of care for children and young people with WS or related conditions. This is done by offering dedicated, specialist outpatient review to families on a yearly or twice yearly basis.

The aims of this service are:

1. To provide a clinical resource and expert advice for medical teams who need information and guidance on WS and its complications.

2. To offer annual assessments of growth and development for affected children, with investigations and advice on management of complications such as diabetes mellitus, diabetes insipidus and visual impairment.

3. To coordinate local healthcare teams, offer advice on disability entitlements, liaise with schools and colleges, providing them with up to date information about WS and patient needs.

4. To provide an opportunity for families to meet, share experiences and offer each other emotional support. To promote audit and research into the best therapies to try to prevent or delay the complications occurring.

5. To offer educational and interactive support for children and their families through learning and teaching opportunities, and group activities

6. To maintain and develop the child and family friendly approach of the service.

7. Access the national genetic testing service with testing for relevant genes.

8. Establish a disease specific centre of excellence and expertise with outreach clinics.

In addition, we also aim to provide an opportunity for families to meet and share experiences. The service will also promote audit and research into best therapies to prevent or delay the complications occurring.

We aim to work closely with Wolfram Syndrome UK and be in close communication with your local hospital and community teams.

What to expect at clinic

Your child may be seen at the specialist Wolfram clinic every eighteen months, where you will be invited to spend two days at Birmingham Children's Hospital having various tests and meeting with several health professionals.

On day one breakfast, lunch and evening meal will be provided.

On the second day breakfast and lunch are provided, you will be at the hospital until approximately 4.30 on the second day so please do not arrange transport home until then.

During these clinics a number of investigations will be carried out. Your child will be weighed, measured, have their blood pressure and HbA1c taken.

Your child will be seen by a Paediatric Consultant Endocrinologist and Diabetes Specialist Nurse for a review which will give you the opportunity to ask any questions you may have.

A genetic consultant or nurse counsellor will take a family history from you, and discuss the genetics of WS with you. At this time, you may be asked to give a blood sample for DNA and gene testing.

You will be having a urodynamics test and the Urologist will discuss the results of the test with you.

The Neurologist will perform a full examination and arrange a brain scan (MRI brain), which will take approximately 60 minutes. He will discuss the neurological aspects of WS.

The Ophthalmologist (if required) will discuss any eye problems. Your child will have their vision and visual field checked and may have test such as Fundus photograph, Retinal nerve fibre layer analysis (RNFLA) by optical coherence tomography (OCT). You will be in the eye department for some time having these tests carried out. If required hearing test will be arranged.

The psychologist will talk to you about your worries or concerns around living with WS, you can meet the psychologist as a family or individual.

The transition coordinator role is to support young people with developing their knowledge and skills about their healthcare. They will offer support and advice with areas such as home life, education, careers or benefits.

The also guide the young person and their family through transfer to adult services.

Play Specialist: If your child finds blood tests and other hospital procedures difficult, we can arrange to have a play specialist present to help prepare and enable your child to understand their treatment and distract them during any procedures to help them cope with their anxieties and fears.

The Wolfram Syndrome UK family support coordinator will talk to you about any concerns you have and will arrange accommodation and other logistics outside the clinic. You do not need to worry about appointment times as they will ensure you attend all of the appointments. You might receive other clinic appointment letters but please do not worry as these will be coordinated for you.

For more information about the annual conference, clinics or just to find out how the family support coordinator can help you, please contact them on:

Tel: 07592 629813

Email: georginaking@wolframsyndrome.co.uk

Web: www.wolframsyndrome.co.uk/contact/

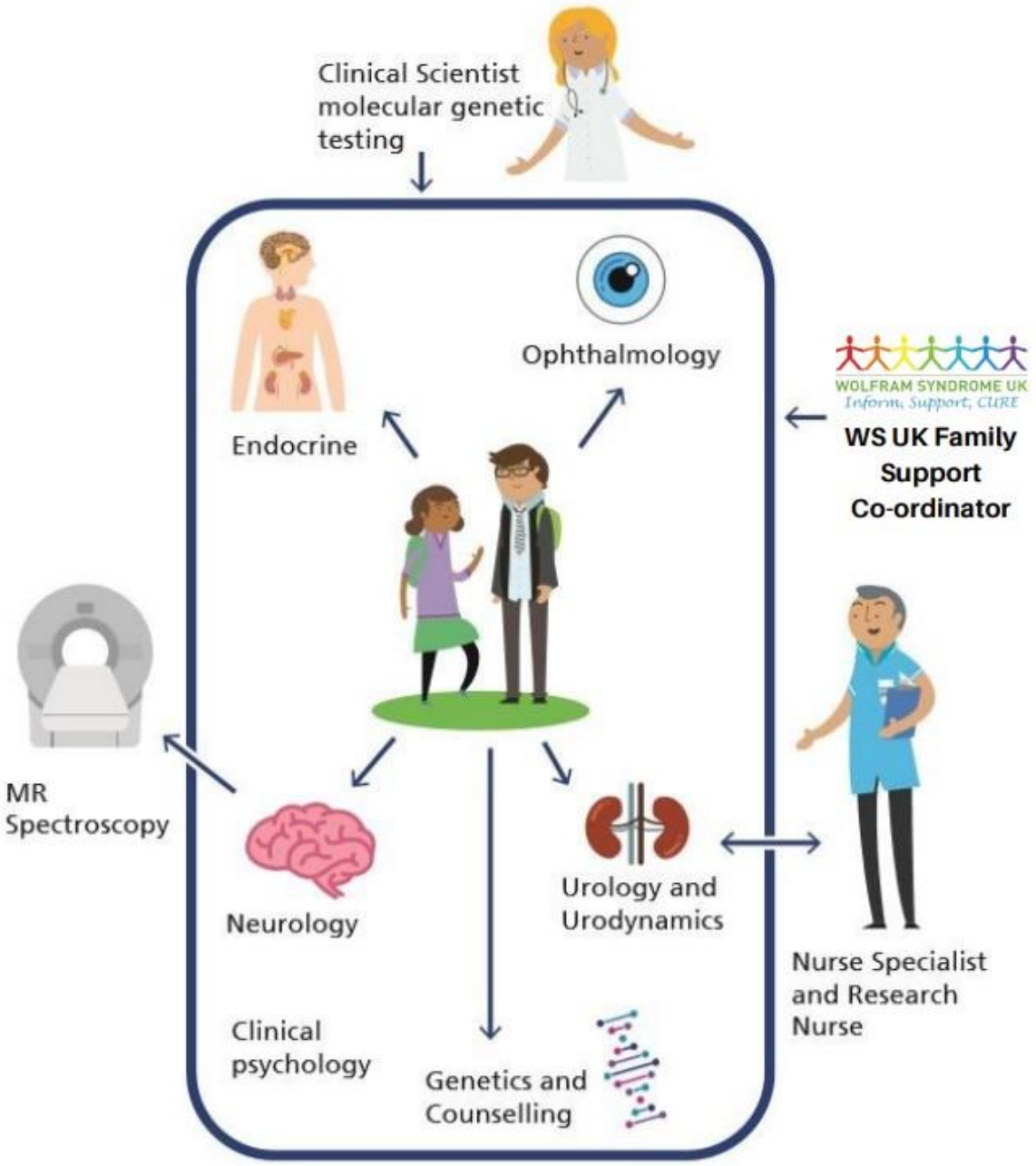
A Clinical Research Nurse will talk with you about research studies that are going on with WS and will answer any questions you may have about these.

Please bring an early morning urine sample with you (a bottle will be provided).

We will all meet up over lunch on the two days so that you can ask any questions and get to know the other families.

Following your appointment; information and test results will be sent to your local health care team to ensure your child receives optimum care locally. You will also receive a letter outlining your consultation with each clinician.

The Wolfram MDT Clinic



Research into WS

Here at Birmingham Children's Hospital, we are conducting research projects to try and find out more about WS.

Before attending clinic, you may be sent some information regarding a research study currently being undertaken at Birmingham Children's Hospital called Euro WABB; An EU Rare Disease Registry. The aim of this research project is to try and learn more about WS along with other rare diabetes syndromes. Whilst at the clinic, if you are eligible to take part in the study, one of the research nurses will come and speak to you about the project and give you a little bit more information and answer any questions you may have. If you would not like to take part and do not wish to discuss the project any further that is fine.

As part of the project, we are also taking skin samples to learn more about WS. Whilst we are talking to you about the study we can go through this procedure in more detail if this is something you would like to take part in. However, it is still possible to take part in the Euro-Wabb project without having a skin biopsy taken. If you would like to have a skin biopsy taken, we will arrange a time during clinic for this to be done by one of the research nurses.

You will see the research nurses throughout the day whilst at clinic. Please feel free to come and speak to us at any point if you have any questions you would like answering.

Blood tests explained

It will be necessary for your child to have regular blood tests to monitor their condition, so that we can advise the best treatments. Below is an explanation of some of the blood tests that your child may have.

HbA1c – Reflects blood glucose control over the last 2-3 months.

Fasting glucose – Glucose levels taken in the morning before eating.

Sodium – Sodium is both an electrolyte and mineral. It helps keep the water (the amount of fluid inside and outside the body cell's) and electrolyte balance of the body. Sodium is also important in how nerves and muscles work.

Potassium – This mineral is essential for relaying nerve impulses, maintaining muscle function and regulating heart beats.

Urea – Reflects how well the kidneys are working.

Creatinine – The blood Creatinine shows how well your kidneys are working.

Blood Osmolality – Measure's the body's electrolyte – water balance. Used to test for Diabetes Insipidus

Thyroid stimulating hormone (TSH) - This test measures the amount of (TSH) in the blood. TSH tells the thyroid gland to make and release thyroid hormones into the blood which is essential to help maintain the body's metabolic rate, heart and digestive functions, muscle control, brain development and maintenance of bones.

Free thyroxine – This test tells us how much thyroid hormone is free in the blood stream to work on the blood.

Follicle stimulating hormone (FSH) – Plays an important part in sexual development. This test measures the level of this hormone in the blood.

Luteinising Hormone – Also plays an important part in sexual development, because LH and FSH work closely with each other these tests are performed together. Taken together the results can provide a more complete picture of your child's sexual maturity and the well-being of the glands that produce these hormones.

A urine test will also be requested for:

Urine osmolality – This is a measure of urine concentration. The results of this test are usually used along with other results to give a picture of the body's fluid balance. Urine osmolality can be used to help diagnose diabetes Insipidus.

Investigations explained

Urodynamic – You will be asked to record several days urinary flow rate at home before the clinic.

When you arrive on the unit you will be met by the nurse who will explain the test to you and you will be shown the flow rate toilet.

When you are ready to pass urine, you will pass urine into the flow rate toilet.

The computer will register the flow and will produce a print out. After each flow rate you will have a bladder scan to check your bladder is empty, this involves putting cold jelly on the tummy and using a probe to create a picture. This is painless.

You may have to repeat the flow rate and scan once or twice more.

Magnetic Resonance Imaging (MRI) - Uses a strong magnet and radio waves to take detailed pictures of your insides.

MRI can give doctors detailed pictures of nearly every part of the body, but they are usually used to help them get a better look at your brain, pelvis, spinal cord and liver. MRI is also good for showing things like muscles, joints, bone marrow, blood vessels and nerves, and organs like your brain and heart.

It is better than having an x-ray because it gives more detail and can show things like swelling, inflammation and blood flow. The pictures taken by MRI are usually two-dimensional, but sometimes a three-dimensional image can be built up and shown on a computer screen.

Why do I need one?

Why do I need one?

Your doctors can see lots about how your brain works from an MRI because it can measure changes in blood flow.

How does it work?

Your body is made up of billions of tiny things called protons. These protons are so small, that they are even smaller than a grain of sand. An MRI works by taking pictures of your protons.

Do I need to do anything before I have the scan?

You must remove any metal items that you are wearing or have on you. You should also make sure you don't have any clothes that have metal zips, fasteners, buttons belts or buckles. Make sure you inform the radiographer if you have an insulin pump, if you wear hearing aids you may be asked to remove them. If you use a CGM this will need to be removed, so bring a spare with you.

If you have a cochlear implant, you will not be able to have a MRI scan.

What happens?

You lie on a bed in a machine called an MRI scanner. It's like a short tunnel, which is open at both ends. The bed is motorised and it passes through the scanner. It usually takes up to an hour.

The MRI scanners can be very noisy, due to the magnet being switched on and off. You will be given headphones to block out the noise.

Will it hurt?

No. MRI is totally painless.

Hearing Tests:

Pure tone audiometry – Younger children are shown how to move a toy (for example, putting a peg into a board) each time they hear a sound. Older children are asked to respond to sounds by saying yes or pressing a button. The sounds come through headphones, earphones placed inside your ear, or sometimes through a speaker (when the test is known as sound field audiometry).

Tympanometry – This is not a hearing test; it is used to check how well the moving parts of the middle ear are working. A small ear piece is held gently in the ear canal. A pump causes the pressure of the air in the ear canal to change. The ear drum should move in and out with the change in pressure. The earpiece measures this by checking the sound reflected by the eardrum.

Eye Screening: The eye department aim to perform a vision test using letters or pictures.

This usually depends on the child's age and co-operation. They also aim to perform: • Vision aid assessment particularly regarding glasses • Vision near and distance • Visual field test • Colour vision assessment.

Investigations also include:

Fundus photograph:

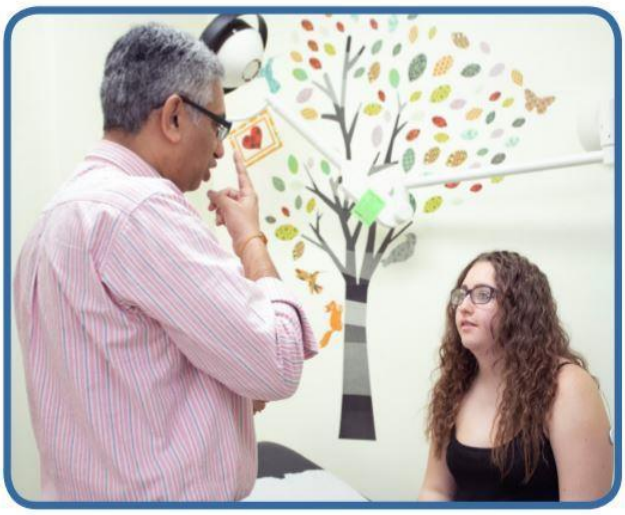
The eye is a little bit like a camera. At the front of the eye are the cornea and the lens that focus the light onto the back of the eye, the retina is a bit like the film or sensor of a digital camera. At the retina the picture is changed into a nerve signal which passes to the brain along the optic nerve. The macula area of the retina is the most sensitive part where we see our most detailed vision.

The photograph of the fundus is taken with a specialised camera. By taking a digital fundus photograph image of the back of the eye (the retina) the doctor can detect and monitor the health of your eyes. The screening picture that is produced captures a clear view of the optic nerve, macula and main blood vessels. Your eyes may be dilated before the procedure, widening (dilating) the pupil increases the angle of observation. This allows the clinician to image a much greater area and have a clearer view of the back of the eye.

You will be asked to sit at the fundus camera with your chin in a chin rest and forehead against the bar. An ophthalmic photographer focuses and aligns the fundus camera. A flash fires as the photographer presses the shutter release, creating a fundus photograph.

Retinal nerve fibre layer analysis (RNFLA) by Optical coherence tomography (OCT) – The retinal scan uses a low intensity light source and a delicate sensor to scan the pattern of blood vessels at the back of the retina, a pattern unique to each individual. It also

measures the thickness of the retinal nerve fibre layer and documents the appearance of the optic nerve head.



For this test to be undertaken you need to be able to sit still fixing on the light inside the camera (green light OCT (blue light).

We may not get all these tests at any one visit however the doctor can also examine the fundus.

Medicines

Your child may be taking a variety of different medicines; you may find it helpful to keep a record of the different medicines and the doses.

Name of medicine	Date started		Date changed		Date changed		Date changed		Date changed	
	Dose	How often	Dose	How often	Dose	How often	Dose	How often	Dose	How often
EXAMPLE Thyroxine	Date: 3.2.17		Date: 17.9.17		Date: 21.3.17		Date:		Date:	
	50 mcgs	Once daily	75 mcgs	Once daily	100 mcgs	Once daily				
	Date:		Date:		Date:		Date:		Date:	
	Date:		Date:		Date:		Date:		Date:	
	Date:		Date:		Date:		Date:		Date:	
	Date:		Date:		Date:		Date:		Date:	
	Date:		Date:		Date:		Date:		Date:	
	Date:		Date:		Date:		Date:		Date:	
	Date:		Date:		Date:		Date:		Date:	

Key

OD
once daily

BD
twice daily

TDS
three times daily

QDS
Four times daily

Nocte
at night

What is transition?

You may have heard the word ‘transition’ being used by health professionals. It is often used as a way to describe growing up and managing changes at key points in your life, while living with Wolfram Syndrome. This includes all sorts of things like: changing schools, choosing a career, making new friends or managing your emotions.

It also includes planning with you and preparing you for your eventual transfer to adult services. It is a gradual process that gives you, your family and anyone involved in your care, time to get you ready to move (transfer) to adult services and discuss what healthcare needs you will require.

This will include deciding which services are best for you and where you will receive that care. You will have an active role, helping to determine your readiness and as part of your transition education, you will use a transition check list to identify your skills and knowledge.

You will have an active role, helping to determine your readiness through setting goals and building your confidence. This process will occur slowly across your adolescence (from Latin: adolescere meaning “to grow up”) giving you time to learn new skills and practice them with your healthcare team.

When will transition start?

The timing of your transition will vary depending on when you are ready. It can start around 11 years old but this varies from person to person. Throughout transition your healthcare team will be supporting you for your eventual transfer to adult health services. Transfer to adult services includes assessing how ready you are to

transfer, exchanging information between services and providing you with practical information about adult health care.

What happens in transition?

Transition is basically all about growing up and developing skills to build confidence and independence so that you gradually learn to manage some or all of your own health care and well-being.

What are transition skills?

Transition skills include lots of things like: speaking up for yourself (advocacy) and setting goals. As your confidence and skills gradually improve you may begin to ask more questions or choose to see the doctor/nurse on your own for a short time.

Transition plans

Transition checklists and plans will be used in consultation with your healthcare team and aim to empower you and your parents/ carers throughout the transition journey. It's a way of letting those who are supporting your healthcare know where the gaps are in your knowledge and to identify which skills need more support.

Transfer to adult care

There is no exact time when you will move to adult services, your healthcare team will plan with you and your family, helping you to look at where your future care will be delivered and how this will fit in with your future plans.

As the transition process prepares you for your eventual transfer to an adult hospital you are encouraged to start thinking about questions to ask your healthcare team.

These could include:

- What is the plan for my transition?
- When am I moving to an adult hospital?
- What is different about adult hospitals?

- Can I choose which adult hospital I move to?
- Can I meet the adult staff before I leave children's services?
- Can I visit the adult hospital to look around?
- Are there any young people I can talk to about moving to an adult hospital?
- How will my condition affect my future, such as college, university or work?

We understand that moving away from the healthcare staff that have been looking after you for many years can be scary, but hopefully by getting involved in the transition process, you will feel more confident and happier about the move.

Appointments

The space below can be used to help you keep track of your hospital appointments.

Who you are seeing

Hospital/location

Date/Time

Who you are seeing

Hospital/location

Date/Time

Who you are seeing

Hospital/location

Date/Time

Who you are seeing

Hospital/location

Date/Time

Who you are seeing

Hospital/location

Date/Time

Appointments

Who you are seeing

Hospital/location

Date/Time

Who you are seeing

Hospital/location

Date/Time

Who you are seeing

Hospital/location

Date/Time

Who you are seeing

Hospital/location

Date/Time

Who you are seeing

Hospital/location

Date/Time

Who you are seeing

Hospital/location

Date/Time

Financial Support

What is Disability Living Allowance (DLA) and who can claim it? Following the introduction of Personal Independence Payments (PIP), DLA is a benefit to help support children under 16 with care and mobility needs.

Adults under 65 who have care and mobility needs should apply for Personal Independence Payment (PIP), while those over 65 should claim Attendance Allowance.

You can claim DLA on behalf of your child if they need more care than children of the same age, meaning they need assistance with things like getting dressed or going to the toilet. You can also claim for mobility meanwhile if they cannot walk or follow an unfamiliar route without guidance. You can claim the mobility component of DLA once your child is 3. If it is the higher rate they need (severe walking difficulties), or once they are 5 for the lower rate (they need guidance or supervision when outdoors).

Who is eligible for DLA?

Find out more about eligibility for DLA and DLA rates on the official government website.

DLA is a benefit to help with the extra costs of raising a disabled child. It takes into account your child's mobility and care needs. DLA is not means-tested.

DLA has 2 parts - care and mobility. You may be paid either one, or both. To qualify for either, your child must satisfy the DLA disability tests for 3 months before being paid and be likely to do so for at least 6 months afterwards, unless they're terminally ill.

Your child can receive only one rate from each component.

If your child is awarded DLA, you may qualify for Carer's Allowance and for help from the Motability scheme.

Read more at https://www.scope.org.uk/support/disabled-.9

How do I claim DLA for my child?

You can either:

• Call the Disability Living Allowance helpline and ask for a claim form (DLA1)

• Telephone 08457123456

• Text phone 08457224433

• Download a DLA1 claim pack from Direct Gov

Your claim date will be the date you call and ask for the form, if you complete and return it in the time given. If you download a claim form, it will be the date your completed form is received. Claims can take around 40 days, and there may be an assessment.

Read more at https://www.scope.org.uk/support/disabled

Employment Support Allowance

What is Employment Support Allowance and who is eligible? Employment Support Allowance (ESA), is a benefit which you can receive if your ability to work is limited by disability or poor health. It's intended to provide you with financial support if you are unable to work.

To be eligible for ESA you must be under state pension age, have an illness or disability which prevents or impairs your ability to work, and you must not be in receipt of statutory sick pay, maternity pay or jobseeker's allowance.

Two types of ESA

• **Contributory ESA** - linked to your National Insurance contributions

• **Income-related ESA** - means-tested and taking into account your other income and savings.

You may be entitled to either or a combination of the two.

Work Capability Assessment

The Work Capability Assessment, carried out by Approved Healthcare Professionals (AHP) on behalf of the Department of Work and Pensions (DWP), tests your eligibility. The assessment has two parts, intended to find out if you have a limited capability for work and then work-related activity.

This normally takes place within the first 13 weeks of your ESA claim. You will receive the basic rate of ESA during this time.

Read more at

https://www.scope.org.uk/support/disabled-people/benefits/employment-support-allowance

How to claim ESA

The quickest method of applying for ESA is to call the government's ESA contact number on 0800 055 6688.

Visit the official government webpage for Employment and Support Allowance to find out what details you'll need when making a claim, and to find out about alternative application methods.

Read more at https://www.scope.org.uk/support/disabled-people/benefits/employment-support-allowance

Claiming travel costs for clinic visits

You can claim back travel costs for clinic visits if you are in receipt of one of the following:

- Income Support
- Income related employment support allowance
- Income related jobseeker's allowance
- NHS Tax credit Exemption Certificate

Contact the Wolfram Syndrome UK Family Support Coordinator for further information on claiming back travel costs on 07592 629813

Accommodation

The Wolfram Syndrome UK Family Support Coordinator/ WS Clinic Co-ordinator will arrange accommodation, which is provided free of charge for families attending clinics at Birmingham Children's Hospital.

Further Information

We hope that this information helps you to understand WS and the service we offer at Birmingham children's Hospital. This information has been produced using the latest available evidence. Further details are available on request.

The UK based WS charity provides support for families and individuals living with WS, as well as helping to raise funds to further research into finding a treatment, to provide an annual conference for families and those affected. The WSUK Chief Executive oversees the running of the charity, writes the quarterly newsletters, and ensures the website and Facebook page are kept up to date and can be contacted on 01903 211358.

Email: admin@wolframsyndrome.co.uk or tracylynch@wolframsyndrome.co.uk

Useful websites:

www.wolframsyndrome.co.uk

www.cafamily.org.uk

www.sense.org.uk

www.diabetes.co.uk

www.association-du-syndrome-de-wolfram.org

website for French Wolfram Association providing information and support for families

www.ncbi.nlm.nih.gov/books/NBK4144/)

This article on gene reviews provides a good review of WS.

www.orpha.net

Orpha is an online database of rare diseases and related services provided throughout Europe. It contains information on over 5000 conditions and lists specialised clinics, diagnostic tests, patient organisations, research projects and clinical trials.

www.euro-wabb.org

The Euro-WABB project is a collaboration of doctor's, scientists and patient support groups from all over Europe. This website provides information on WS and other rare genetic forms of diabetes.

Further information and internet access is also available in the Family health information centre at the hospital. Tel 0121 3338505 or email child.infoctr@bch.nhs.uk

Fundraising

Give as you Live brings together thousands of retailers that have signed up to donate to Wolfram Syndrome UK (WSUK) a percentage of every online purchase you make. Just by shopping online with stores including John Lewis, Play.com and Expedia, you can raise as much as £50 and more for WSUK, without adding to the cost of the shopping.

So why wait?

Head to www.giveasyoulive.com/join/wolframsyndrome today and support us every time you fill your basket.

This information has been produced by the WS team within Diabetes Home Care at Birmingham Children's Hospital.

Clinical guidelines for medical staff

Following diagnosis, clinical management of WS requires the coordinated involvement of many different clinical specialties. Together with clinical specialists from WS, Euro-WABB has developed clinical management guidelines for health professionals for WS, helping to ensure that patients receive a timely diagnosis and optimal clinical management and care.

These have been developed through multidisciplinary guideline development meetings, contribution and peer review by international experts in the fields, and advice from family support groups. Reassessment of existing and potential patients from the registry has been used to refine the agreed diagnostic criteria and to facilitate the development of consensus referral, care and management pathways. The development of these guidelines is identifying knowledge gaps and specific learning needs of healthcare professionals.

In partnership with family support groups, educational materials and training tools are being developed to disseminate to target groups such as medical students and primary care professionals. These guidelines are freely available via the website www.euro-wabb.org

Birmingham Women's and Children's NHS Foundation Trust, Steelhouse Lane, Birmingham. B4 6NH Telephone 0121 333 9999 Fax: 0121 333 9998 Website: www.bwc.nhs.uk

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