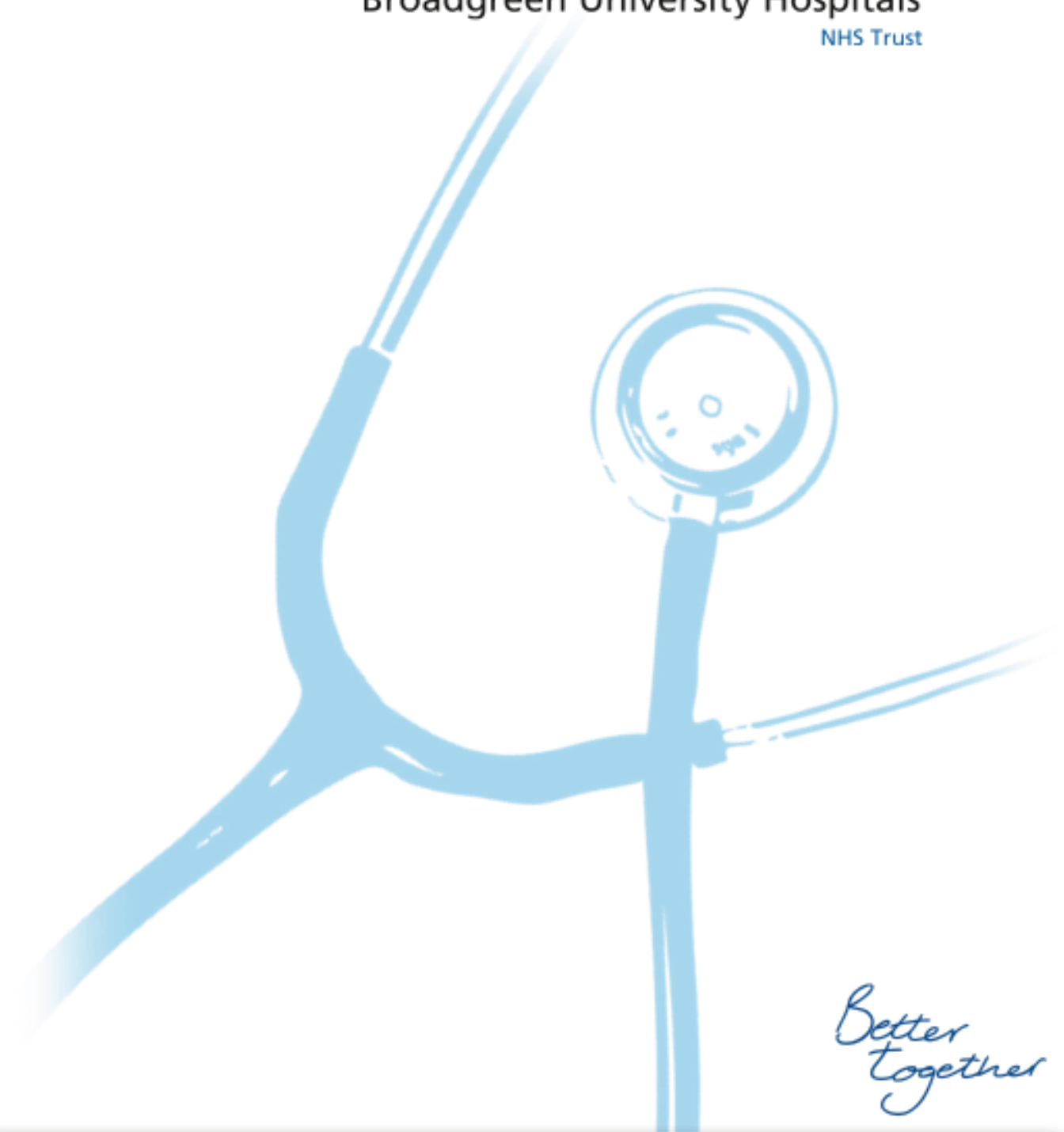


The Royal Liverpool and  
Broadgreen University Hospitals  
NHS Trust



*Better  
Together*

Patient Information

**Robert Gregory National Alkaptonuria  
Centre Booklet**



# Introduction

Alkaptonuria (AKU) is a rare condition causing significant health problems dominated by arthritis in the spine and weight bearing joints.

Because it is rare, people with this condition are dispersed around the country and lacking effective health care.

AKU was first identified and described in 1902 by Archibald Garrod. It is a rare disease affecting one in every 250,000 to one in 1,000,000 people.

Many doctors will never see a patient with Alkaptonuria.

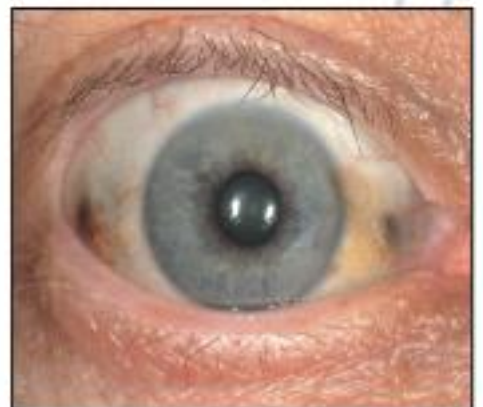
## What Alkaptonuria does

People with AKU do not have enough of an enzyme called homogentisic acid oxidase. The body uses this enzyme to break down a substance called homogentisic acid.

Because normal amounts of this enzyme are missing, homogentisic acid is not used and so builds up in the body.

Some is eliminated in the urine but the rest is deposited in body tissues where it accumulates at 2,000 times the normal rate.

This accumulation causes ochronosis, which is a blue-black discolouration of connective tissue including bone, cartilage and skin caused by deposits of ochre (black) coloured pigment. Homogentisic acid is toxic and its build up in the tissues of the body leads to multiple and chronic health problems for those people with AKU.



## Diagnosis and Symptoms

Babies born with AKU do not suffer any immediate effects. However, their urine will turn a dark colour after several hours' exposure to air because of the presence of homogentisic acid in the urine. Parents may notice dark staining on a baby's nappy.

Many people with Alkaptonuria are not diagnosed until symptoms appear later in life, after years of accumulation of homogentisic acid in their body tissues. The onset of joint symptoms is variable in men and women and generally begins after age of 30. Generally there is increasing joint pain and reduced and painful use of the large weight-bearing joints, knees, hips, spine and shoulders.

### Symptoms may include:

- severe arthritis of the spine and other large joints
- heart valve damage
- stones forming in the kidneys, prostate, gall bladder and salivary glands
- rupturing (bursting or breaking) of tendons, muscles and ligaments
- fractured bones
- hearing problems
- kidney damage

# Robert Gregory National Alkaptonuria Centre

The Robert Gregory Alkaptonuria Centre (NAC) was established by the NHS National Specialised Services Commissioning Group from April 2012 to provide assessments and treatments for patients with alkaptonuria (AKU). It is based at the Royal Liverpool University Hospital.

The Royal Liverpool University Hospital has been researching and managing AKU since 2003. In recognition of the unique expertise that exists in Liverpool, the NHS National Specialised Services has designated the hospital as the centre to host the NAC. The lead clinician at the NAC is Dr L Ranganath.

The NAC is located at:

Ward 9B  
Royal Liverpool University Hospital,  
Prescot Street,  
Liverpool  
L7 8XP, UK

The NAC ward is located on the 9th floor of the hospital with five single rooms set aside for patients with AKU.

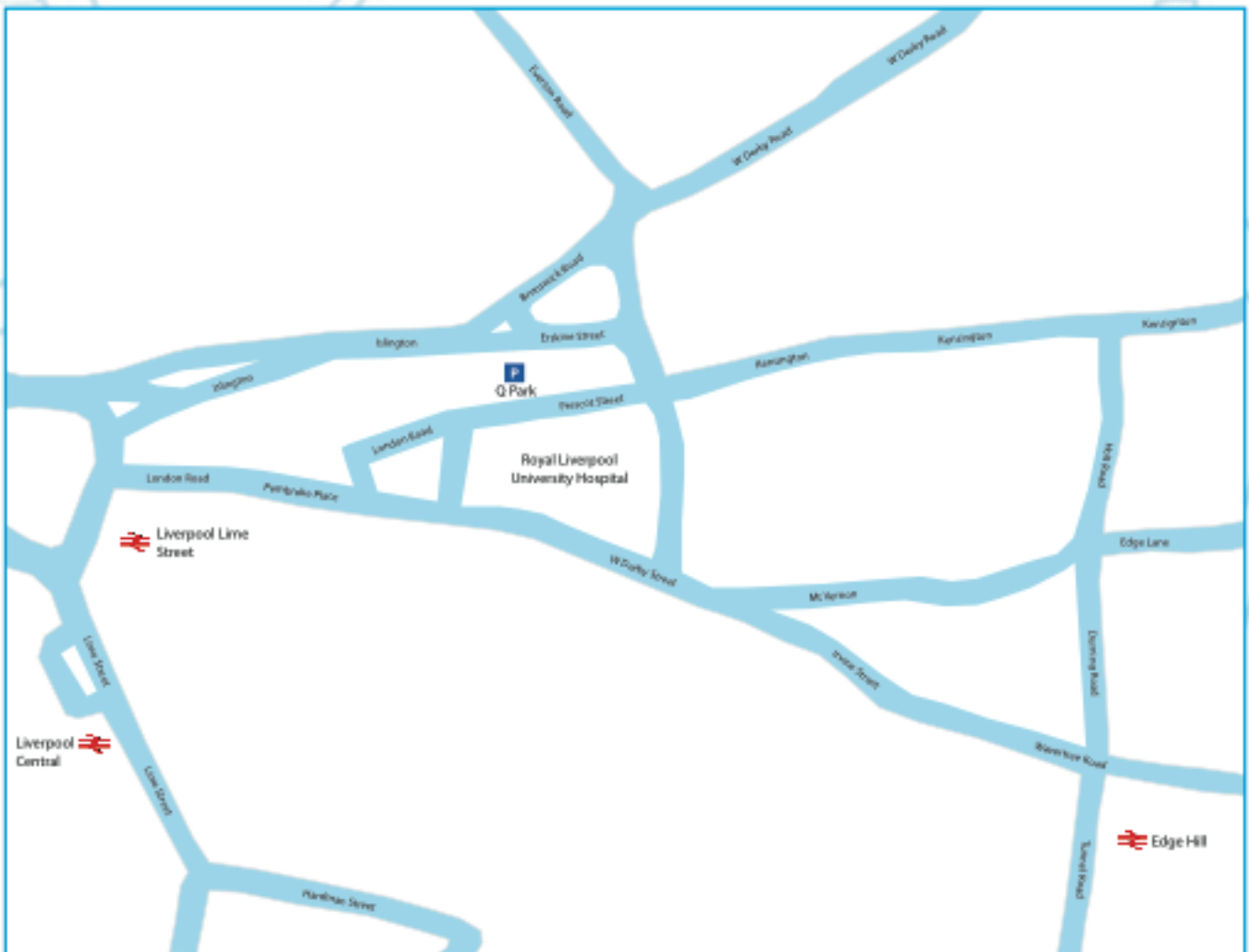
Two of the rooms have ensuite facilities while the other three have access to shower facilities suitable for disabled or wheelchair access. Each room has a TV for which there is a charge. This is done by purchasing a pay to view card from the hospital reception on the ground floor.



## Directions:

The hospital is located in the centre of Liverpool and is well served by local transport networks.

The Royal is less than a mile from Lime Street Rail Station and so is within walking distance, along London Road. Please bear in mind that this is an uphill walk. There are several bus stops serving all directions around the road perimeter of the hospital with others within a short walk. You can also take a taxi from the station to the hospital.



## Cars

The hospital is well signposted throughout the city and is located on a main road. A drop off point is situated at the front of the hospital and car parking facilities in the nearby Q-Park.

## From the M62

The hospital is approximately three miles from the M62 and is signposted.

## From the Mersey Tunnels

Queensway – turn left at the roundabout, at the tunnel exit and immediately bear right into the filter lane and turn right at the traffic lights. Follow signs for the hospital and visitor/disabled parking.

Kingsway – follow the exit slip road onto the main carriageway keeping in the left hand lane. At the traffic lights bear left and follow signs for the hospital and visitor/disabled parking.

Parking on the main hospital site is limited to staff and permit holders. Patients and visitors can park in the nearby secure Q-Park facility, which is situated less than a five minute walk from the hospital and is open 24 hours a day, seven days a week.

The vehicle entrance to Q-Park is on Epworth Street (off Erskine Street). To access this you must follow the signposts through the one way system onto Epworth Street. A pedestrian exit is available on the Prescott Street side of the car park.

### Disabled parking and shuttle bus facility

Disabled car parking is provided at the Q-Park multi-storey. Disabled spaces are available on the entry level and are well marked and signposted. Staff are available to provide assistance to patients and visitors as required.

Q-Park has been assessed by the Trust's Patient Council and Liverpool City Council Disability Discrimination Act officials to ensure it is DDA compliant.

The car park has a designated pick up zone and improved entrance and exit areas including better crossing points outside the car park.

A shuttle bus service will run between Q-Park and the hospital buildings to provide transport assistance to anyone requiring it. The bus runs on request so if the bus is not there as you arrive, please speak to security or car park staff, who will radio the bus for a pick up within a few minutes.

There is a height restriction of 2.1 metres at Q-Park so if you have a vehicle above this height please contact the Trust's Security Team on 0151 706 2228 to make alternative arrangements.

### Q-Park contact details:

Epworth Street, off Erskine Street, Liverpool, L6 1LY  
0870 4420104  
info@q-park.co.uk  
www.q-park.co.uk

Some assistance with the cost of parking may be available from the AKU Society.

There are no overnight facilities for family members or carers wishing to travel to Liverpool with the person attending the centre. However, there are a number of hotels within the local area and the AKU Society can provide details of available options. Please contact Lesley Harrison, Patient Support Officer, for more information.



# Objectives of NAC

## The NAC will:

- Provide a single national centre of expertise for comprehensive assessment of AKU patients.
- Deliver expert one-stop care that will enable monitoring of disease and treatment by an experienced multidisciplinary team.
- Offer and commence effective disease modifying therapy for AKU patients employing nitisinone.
- Allow access to expert surgical and medical assessment.
- Make available an urgent review of health problems if clinically required.
- Formulate integrated, holistic management plans for patients that will be communicated to the local healthcare providers. This will be supported with on-going liaison and expert advice to maximise the extent and efficacy of local clinical management.

## Referral to NAC:

If you are over the age of 16, and have a proven diagnosis of AKU, you are eligible to attend the NAC. Referral will generally be via your GP, and once received by the Royal Liverpool University Hospital will be assessed by Dr L Ranganath. Following this assessment, patients will be invited to attend the NAC.

## Initial Questionnaire:

You will receive a questionnaire prior to your visit. This is to assess your medical history and ensure that the correct support is available on the NAC ward on your arrival. If you need assistance with completing the questionnaire, help is available from the AKU Society.






## Your Visit to the NAC:

The first visit to the NAC will be over four nights and four days followed by yearly visits of three nights and three days. You will need to arrive around lunchtime on the first day, when you will be clerked in. There will be a busy schedule over three and a half days. While many of the tests and investigations will be carried out on the ward, some will require visits to other departments within the hospital. The NAC will provide nitisinone treatment with a low protein diet and physiotherapy. Any potential complications of AKU will be identified and reported to your local healthcare team so treatment between visits can be supplied by your local GP or hospital.

As per the conditions set out for the NAC, you would be expected to have all the assessments and treatments.

These procedures will lead generate a lot of data, which could help us understand AKU better. Such data could be used for research purposes. We could publish results from the study but if we do, we will make sure you cannot be identified in any way. Please let us know when you are visiting ward 9 B that you understand this and have no objection to it.



# Timetable of NAC clinics

An example timetable for your first visit to the NAC is below. Any subsequent visits will depend upon the severity of your condition, but we expect them to be shorter.

| Day 0   |   |                          |                            |                                       |   |   | Night 1                                  |
|---|---|--------------------------|----------------------------|---------------------------------------|---|---|--|
| Arrival in Liverpool (Explanation of NAC/signing consent forms)     |   |                          |                            |                                       |   |   |  |
| Day 1   |   |                          | Day 1                      |                                       |   |   | Night 2                                  |
| 9.00-10.00  | 10.00-11.00                               | 11.00-12.00              | 13.00-14.00                | 14.00-15.00                           | 15.00-16.00                                   | 16.00-17.00                                 |  |
| Patient clerked   | Bloods, Urine, Height, Weight, BP         | Photos, start Nitisinone | H<br>O<br>N<br>Z<br>U<br>L | Eye consult and Tests                 | Cardiology consult and Tests                  | Orthopaedic consult and Tests               | Bone mineral density                     |
| Day 2   |   |                          | Day 2                      |                                       |   |   | Night 3                                  |
| 9.00-10.00  | 10.00-11.00                               | 11.00-12.00              | 13.00-14.00                | 14.00-15.00                           | 15.00-16.00                                   | 16.00-17.00                                 |  |
| Isotope bone scan and extent of arthropathy, random urine and blood | Diet and low protein diet and Psychometry | Hearing and voice        | H<br>O<br>N<br>Z<br>U<br>L | Rheumatology consult and tests and Rx | Physiotherapy consult and Rx                  | Xray spine and other                        | Ultrasound abdomen                       |
| Day 3   |   |                          | Day 3                      |                                       |   |   | Night 4                                  |
| 9.00-10.00  | 10.00-11.00                               | 11.00-12.00              | 13.00-14.00                | 14.00-15.00                           | 15.00-16.00                                   | 16.00-17.00                                 |  |
| Target MRI scan random urine and blood (Genetic studies)            | Spinal consult and ear biopsy             | Questionnaires           | H<br>O<br>N<br>Z<br>U<br>L | Questionnaires                        | Specific problem solving (pain/orth op/spine) | Patient education and summing up Nitisinone | Discharge (blood/urine) Discharge letter |

# Additional Information



## Nitisinone therapy:

Nitisinone is a drug not licensed for use in AKU. There is good evidence that it may work as a treatment for AKU. Through the NAC, nitisinone will be made available to you, following the complete medical examination during your visit. When taking nitisinone, it is recommended that you follow a low protein diet, and a dietician will meet with you to provide advice.



When you leave the NAC, you will be prescribed with six months supply of nitisinone - followed by a further six months supply that will be sent directly to your home. You will also be given a nitisinone information booklet to explain what dosage is required and when to take it, how to store nitisinone, and information on any possible side effects.

Treatments will be monitored by regular assessments during visits and by phone calls in between annual visits. If you have any concerns, please contact the NAC team (see further information, on back page).



## Consent forms:

There is no need to sign a consent form for the assessment and treatments since this is part of the standard service you will receive when you are at the NAC. However, you will have to send a standard consent form for the medical photographs to be taken during your visit to the NAC. This will be further explained to you at the initial clerking session on your first day at the NAC.

### Dietary Advice:

Low protein diets by themselves are not effective to prevent progression of AKU. However, low protein diets will make nitisinone treatment safer, by reducing the availability of tyrosine, the precursor of homogentisic acid. After nitisinone use, tyrosine levels generally increase and a low protein diet will minimise the rise in tyrosine. A separate nitisinone information booklet will be provided at the NAC to explain all about the drug in more detail.

### Discharge from NAC:

On completion of the programme of assessments and treatments at each visit, you will be discharged to your local area Health Trust. If any health problems develop in between annual visits, and they cannot be dealt with locally, you will be able to attend Liverpool for additional assessment and treatment that falls within the scope of the nationally commissioned service.

### Reports:

On completion of your NAC clinic, you will receive a discharge summary letter. This is a detailed comprehensive report on the totality of all assessments and treatments and guidance for further local monitoring and treatment and will be sent to you 10 working days after being discharged. This report will be sent to you, your GP and any other named consultants or therapists actively involved in your care. You will be asked to provide your consent for this communication.

### Role of the AKU Society:

The AKU Society, the charity for AKU patients will be providing additional support alongside the NAC, including:

- Assistance with travel, accommodation and other logistics, working with the hospital's appointments service and liaising with local services for referrals and co-ordination
- Meeting with patients during the NAC clinics
- Emotional support and care to patients and family, carers both during the NAC clinics and between visits.
- Publicising the existence of the service to local GPs and other medical professionals.



### Monitoring and evaluating the NAC from the patient perspective:

The AKU Society will work with the RLUH to review and improve the NAC on an annual basis through patient and clinician surveys and interviews.

### Safety Group:

There is a risk management group that includes Royal Liverpool University Hospital (Dr L Ranganath, Dr T Kennedy, Mr Craig Evans and Professor S Vinjamuri) and AKU Society (Dr N Sireau). The NAC will report to the Department of Health annually.

### About the AKU Society:

The AKU Society is a charity that supports AKU patients and funds research into finding treatments. Set up in 2003, it has a strong partnership with the Royal Liverpool and Broadgreen University Hospitals NHS Trust. It is also at the centre of a fast growing international network of AKU Societies and research centres spanning Europe, North America, the Middle East and Asia.

### About Robert Gregory:

Robert Gregory is an AKU patient and co-founder of the AKU Society along with Dr L Ranganath. When Robert realised that little was being done for AKU patients, he decided to set up the AKU society as the world's first charity exclusively dedicated to the disease. The National AKU Centre has been named after Robert in recognition of his key role in driving forward the AKU movement.



## Further information:

### Dr L Ranganath

Consultant, Royal Liverpool University Hospital  
Tel: 0151 706 4197  
Email: lrang@liv.ac.uk

### Lesley Harrison

Patient Support Officer, AKU Society  
Tel: 07586 759 028  
Email: lesley@akusociety.org.

### Lyndsey Kinsella

RLUH NAC Co-ordinator  
Tel: 0151 706 3523  
Email: Lyndsey.Kinsella@rlbuht.nhs.uk

### AKU Society Office

66 Devonshire Road  
Cambridge  
CB1 2BL  
Tel: 01223 322 897  
Website: www.alkaptonuria.info

### Oliver Timmis

Communications Manager, AKU Society  
Tel: 07799 037 726  
Email: oliver@akusociety.org

## Author:

**Review date:** August 2015

All Trust approved information is available on request in alternative formats, including other languages, easy read, large print, audio, Braille, moon and electronically.

يمكن توفير جميع المعلومات المتعلقة بالمرضى الموافق عليهم من قبل انتمان المستشفى عند الطلب بصيغ أخرى، بما في ذلك لغات أخرى وبطرق تسهل قراءتها وبالحروف الطباعية الكبيرة وبالصوت وبطريقة برايل للمكفوفين وبطريقة مون والكترونياً.

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Dhammaan warbixinta bukaanleyda ee Ururka ee la oggol yahay waxaa marka la codsado lagu heli karaa nuskhado kale, sida luqado kale, akhris fudud, far waaweyn, dhegeysi, farta braille ee dadka indhaha la', Moon iyo nidaam eletaroonig ah.

in partnership with

**AKU**

• alkaptonuria society •

The Royal Liverpool University Hospital NHS Trust  
Prescot Street  
Liverpool  
L7 8XP

Tel: 0151 706 2000  
Web: [www.rlbuht.nhs.uk](http://www.rlbuht.nhs.uk)

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