

Ataxia Telangiectasia

A patient's guide

At Papworth Hospital we aim to run a personalised service to manage all the problems encountered by patients with Ataxia Telangiectasia (AT). We work as a multi-disciplinary team to allow us to deliver the best care.

In order to improve the service, we welcome feedback from patients and as part of this process it was highlighted that a patient information leaflet would be helpful, outlining what to expect of your visit to Papworth Hospital and the nature of your assessments. We hope this leaflet contains all the information you require to understand the nature of your assessment at Papworth Hospital, the tests performed and the reasons for them.

Papworth Hospital

Papworth Hospital is located approximately eight miles north-west of Cambridge in the village of Papworth Everard. The National Adult Ataxia Telangiectasia service is commissioned here because it was recognised that we have the facilities and expertise to provide the best care.

The service is hosted by the Respiratory Support and Sleep Centre (RSSC) at Papworth Hospital. The RSSC has extensive experience in the care and management of patients with neurological disorders affecting the respiratory system. For travel and other information about the RSSC please visit: www.papworthrssc.nhs.uk

Referral

Patients may be referred by other specialists or GPs but it is anticipated that the majority of patients will transition from the paediatric services provided by Nottingham City Hospital.

Patients are admitted on a Sunday with planned discharge on Tuesday (a two night stay). Patients are provided with their own room with ensuite facilities and space for a relative or carer to sleep overnight. The rooms are part of the RSSC ward and have 24/7 nursing provision.

Each room has a television but unfortunately we do not have wifi. All meals will be provided. It is helpful if you

bring all your medication with you. The reason for admission is to enable all the tests and assessments to occur in a timely fashion whilst recognising the difficulties that patients with AT may have with these processes. Often help with transport and travel arrangements is available from the AT Society.

Admission

On arrival at the hospital you will be admitted by the nursing and medical team. This involves taking a history of your medical problems and examining your heart, lungs, abdomen and nervous system.

As part of the evaluation of your lungs a blood gas (a type of blood test) will be performed to measure the levels of oxygen and carbon dioxide in your blood accurately. A recording of the oxygen and carbon dioxide levels in the blood will be performed overnight. This involves a peg being placed on one finger that uses light to measure oxygen levels and a small warm patch placed on the forearm to measure carbon

dioxide levels.

During the course of the following two days you will have a number of assessments and tests as laid out in the enclosed schedule. Following is an explanation of the nature and reason for these tests and assessments.

Tests

1. Pulmonary function tests

These assess how well your lungs are able to move air in and out of the chest, how well your lungs absorb oxygen and the strength of your breathing muscles. This involves a variety of blowing tests. Our staff are highly sympathetic to the problems you may have with these tests and will understand if you are unable to complete some or all of these assessments. We plan to repeat these tests, if possible, each time you are seen to enable us to establish if your AT is causing respiratory problems and if these are getting better or worse.

2. Chest CT scan

All new patients to the Adult Ataxia Telangiectasia service

are offered a chest CT scan as part of their initial assessment. The team is sensitive to concerns about radiation risks and is happy to discuss this. No examinations will be carried out without the patient's consent. The CT scan is performed to identify all the issues AT may cause with the lungs and is reviewed by our radiologists who are experts in chest conditions.

We know that AT is a disorder associated with increased sensitivity to radiation at levels associated with radiotherapy, rather than those associated with diagnostic studies such as CT. Discussions with AT clinical experts at the international meeting in October 2004, at the Nottingham AT Clinic, with Professor Malcolm Taylor, University of Birmingham and with the AT Society have produced a consensus that a high-resolution CT chest scan is a low risk procedure and important for clinical assessment in AT. A chest CT scan would not be repeated routinely after this but would be performed if there were clinical concerns.

The test will involve you lying as flat as possible on a special bed that is then passed through the CT scanner. The CT scanner is a bit like a giant 'polo' mint and you pass through the centre on the bed. The scanning process is fast and takes from 30 to 60 seconds. The CT scanner is not very deep (1.5 to 2 feet) enabling you to see out of either end at all times, reducing any sensation of claustrophobia.

3. Videofluoroscopy

Videofluoroscopy is a test to assess your swallowing. Our clinical experience to date has shown that people with AT can be unaware that food and fluid may be 'going down the wrong way' in to the lungs when they swallow. This can cause major problems with the lungs and infection. Appropriate therapist input can reduce the risk of these problems greatly. The test does involve some exposure to radiation but at a low dose. The test would not be repeated routinely but would be performed if there were clinical concerns.

The test is co-ordinated by our

speech and language therapist. You will be asked to eat and drink a variety of consistencies of food and fluid that contain a special dye that shows up on X-rays. You will be filmed from the side using a special X-ray camera so we can see what happens to the things you swallow.

4. Blood tests

You will have blood taken from your arm to assess your immune system and to ensure you are not anaemic or have other problems with your health.

Assessments

1. Respiratory physician

Respiratory problems can be a particular issue for patients with AT. The combination of reduced efficiency of the immune system, poor ability to cough and swallowing difficulties can lead to frequent chest infections. Damage to the lungs might occur making future infection more likely. People with AT may under-breathe due to problems with muscle co-ordination and strength. The aim of the respiratory assessment is to

identify any problems and provide you with the right advice and treatment to manage them.

You will be seen by a consultant respiratory physician with a specialist interest in AT and the problems it causes. The consultant will take a history, examine you and review all your test results. The issues raised and the best ways to deal with them will be discussed with you.

2. Neurologist

Neurological symptoms are one of the hallmark features of AT and are often the main cause of disability. Most patients with AT experience progressive unsteadiness and clumsiness but other symptoms can also occur and vary significantly between individuals. These include muscle spasms, jerks, involuntary movements, abnormal sensations, numbness or weakness. Furthermore, visual symptoms are common including difficulties with tracking, jumpy vision or double vision.

During your stay at Papworth

Hospital, you will be assessed by a consultant neurologist with a special interest in AT. The consultant will conduct a detailed clinical examination in order to determine your current neurological problems and symptoms. You will be given advice on how to manage certain neurological symptoms, for example pain, spasticity, abnormal movements or tremor. Furthermore, the assessments are useful in order to determine more formally your individual disease progression.

3. Immunologist

Weakness of the immune system may occur in patients with AT. This can take a number of forms and affects your body's ability to resist infection. Recurrent infections are unpleasant, may lead to hospitalisation and can cause damage to the organs affected. Usually in the presence of immune weakness it is possible to define the problem and to provide suitable treatment to limit the impact this weakness has. Depending on your symptoms and test findings, treatments such as prophylactic

antibiotics, immunisations or possibly immunoglobulin replacement therapy may be organised.

4. Physiotherapist

The specialist physiotherapy team aims to assess your individual needs and provide tailored advice that may include exercise regimes and some techniques to improve your ability to cough.

The physiotherapy team liaises closely with the occupational therapist in order to assess the practicality and safeness of how you manage activities at home and in general. The aim of our team is to reduce complications such as muscle contractures, maximise physical function and to reduce the risk of complications such as chest infections. Often this will involve trying various techniques to see which one is most effective for you. Further input and follow-up in the community may be helpful and will be arranged as required.

5. Occupational therapist

Occupational therapy aims to provide the means to maximise

your ability to manage everyday activities. These activities include accessing your accommodation and the community in which you live and also details such as getting on and off the bed, toilet and chair, managing personal care and considering your role within society. The symptoms of AT can have a significant impact on managing these every day activities and potentially on the quality of your life.

You will have an opportunity to meet with an occupational therapist who has a special interest in AT. Together we will review details of your accommodation, functional mobility and lifestyle. Any problem areas which may benefit from further input will be identified and an action plan agreed.

We may be able to address some issues immediately whilst others will require research and onward referral to local teams. It has been found that the support from Papworth Hospital's specialist team can help interventions to be

correctly prioritised. It is helpful if you can complete the self-assessment questionnaire before coming for assessment as it helps identify areas for discussion.

6. Speech and language therapist

The speech and language team will assess you and attempt to address communication and swallowing issues. Advice and exercises to help improve communication are provided and a problem solving approach applied. The therapist will assess your ability to swallow using questionnaires, direct observation and, if necessary, video fluoroscopy. We will offer advice on methods to improve your ability to swallow and to reduce the risk of aspiration.

7. Dietician

Input from a dietician is important in patients with AT in order to ensure they receive the correct nutrients and sufficient calories to maintain and promote health. The dietician will run through your usual diet and assess your nutritional status and

requirements. If needed, suitable supplements may be prescribed and local follow-up in the community organised. Occasionally, due to swallowing or other problems associated with AT, it may be necessary to discuss with you and inform you of alternative feeding methods, such as PEG tubes, in order to allow you to make an informed decision about your future care.

8. Social worker

Social workers are an integral part of the AT assessment and review process. They will meet with you on your own or with your family or carers, according to your wishes. We will ask about background and general day-to-day circumstances, but you are free to tell us as much or as little as you wish.

Most patients have contact already with their local Social Services Department and other care agencies, but we can make referrals, if necessary, and liaise with them over any issues that you may have. We do not have the power to change local policy or procedures, but we can offer advice on care

services in general. In addition we can offer help and advice on benefits, employment issues, housing, and we can liaise with other relevant organisations, as necessary.

Our contact with AT patients is not confined to those occasions when they visit Papworth Hospital. We are happy to communicate with patients and/or their family or carers by telephone, email or letter.

What happens next?

You will find that the time you spend in the hospital will be quite busy and it may be difficult to absorb all the results and you are advice given.

Your case will be discussed at our multi-disciplinary meeting and a written report explaining the outcome of your tests, assessments and recommendations for future care will be sent to you, your GP and any other relevant healthcare professionals involved in your care. If there is anything you do not understand, please feel free to contact us so that we can explain things in more detail.

You will be offered annual follow-up for assessment but we would be keen to see you sooner if there were any new problems. We are available to provide advice and support as necessary to healthcare professionals involved in your care. If you choose, we are happy to communicate your results and other aspects of your care to the AT Society.

Everybody with AT has a unique set of problems. The aim of our assessment is to address all of your issues in detail. If you feel anything has not been addressed or could be

performed to a higher standard we would be grateful if this information could be fed back to us directly or via the AT society if you prefer.

We are a research active unit and you may be asked to take part in any ongoing projects. This is entirely on a voluntary basis and if you choose not to take part it will not affect the care or treatment you receive. Our aim is to provide you with a high quality service that addresses your problems and we hope, with research and education, improve the quality of life of other adults with AT.

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