TSC and kidneys
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Introduction

Tuberous sclerosis complex (TSC) is a rare genetic condition that causes tumours to develop in different parts of the body. These tumours most commonly occur in the brain, eyes, heart, kidney, skin and lungs. They are benign (non-cancerous) but can cause a range of associated health conditions and complications.

About 80% of people with TSC will have renal (kidney) AMLs (angiomyolipomas), but only about half of these people will experience serious health problems. The rest will need surveillance but eventually they will not need treatment. Kidney problems are not common in early childhood but become more common as individuals get older.

This leaflet describes the kidney problems that people with TSC may develop and what treatments are available.
Monitoring

The kidneys of a person with TSC should be scanned (by ultrasound, CT or MRI) at the time of diagnosis and then at one- to three-year intervals if no cysts or angiomyolipomas (AMLs) are found. MRI scans are usually the best choice, because important problems can be missed if someone only has ultrasounds.

This pattern of monitoring continues throughout adulthood even if the images at adolescence are normal. The monitoring of blood pressure and blood tests for kidney function is also recommended once a year.

If growths are found, a follow-up scan should be repeated every six to 24 months, unless symptoms develop or there is an unusual growth pattern. If this occurs, further investigations and treatment may be needed.

About the scans

**Ultrasound:** Sometimes called a sonogram, this is a procedure that uses high-frequency sound waves to create an image of part of the inside of the body.

**Computed tomography (CT):** A type of imaging that uses x-rays to create a computer-generated, 3D image of a body structure.

**Magnetic resonance imaging (MRI):** MRI uses a powerful magnet, radio waves and a computer to produce images of the inside the body.
Renal (kidney) AMLs

AMLs are tumours made up of fat, muscle cells and abnormal blood vessels. It is thought that AMLs begin to grow very early in childhood and that they continue to grow and increase in number over time. About 80% of adults with TSC have AMLs in both kidneys. Whether or not the AMLs cause problems that need treatment will depend on their size, rate of growth and where in the kidney they are.

AMLs are usually slow growing. They can affect kidney function and obstruct the flow of urine. About 40% of people with TSC develop reduced kidney function by middle age. It is not clear whether this is mainly due to AMLs, or another direct effect of TSC. But it usually occurs in those who have bilateral (on both sides) large AMLs.

AMLs in the kidneys can also lead to increased blood pressure. Controlling this high blood pressure with medication is very important, because if it is left untreated it can speed up the loss of kidney function and lead to an increased risk of strokes and heart attacks in the long term.

The blood vessels within AMLs are abnormal and can develop weak spots in their walls (called aneurysms) that can burst. If this happens the bleeding will show up in the person’s urine.

AMLs vary in size from 1mm up to more than 20cm in diameter. The bigger the AML and the more blood vessels it has, the more likely it is to need treatment to prevent problems. Once an AML has reached 3cm in diameter, and is still growing, it is likely treatment will be needed in order to avoid complications.
Treatment for AMLs that are causing problems

In cases where the AMLs are causing immediate problems, such as a haemorrhage (bleeding), an immediate procedure may be needed. Treatment options include:

- **Embolisation:** This is the deliberate blocking of the blood flow to the AML tumour by the placement of a “blocking agent” into the blood vessel that feeds it. The doctor makes a small cut (usually under a local anaesthetic) in the groin and inserts a catheter (long, fine plastic tube) into a blood vessel. The catheter is then moved into position using x-ray pictures on a screen. The blocking agent is injected through the catheter into the selected blood vessel and this stops the bleeding and causes the AML to shrink.

- **Surgery:** It may be possible to remove the problematic AML surgically. In some cases a whole kidney may have to be removed but this is avoided if at all possible.

- **Dialysis and transplantation:** For the small number of individuals (about 1%) who go on to experience kidney failure, dialysis and transplantation may be an option.

For those who are at risk of complications but who do not require immediate surgery, the medication Everolimus (Votubia®) is now the recommended treatment.

- **Medication:**
  Everolimus belongs to a group of drugs known as mTOR inhibitors, which are effective at shrinking some growths caused by TSC. Everolimus is now funded for use in children aged over three with AMLs larger than 3cm and growing, as well as being licensed for adults.
Risk factors warranting treatment with Everolimus include the following:

- Tumours which are growing and larger than 3cm in diameter.
- The presence of aneurysms.
- The presence of multiple tumours or tumours in both kidneys, with one 3cm or larger.

The best treatment for any individual needs to be carefully considered by a TSC kidney expert. Before making a treatment recommendation, the specialist will take into account the exact nature of the problems the patient is experiencing and all their other circumstances.

Renal (kidney) cysts

Renal cysts may be present at birth and in younger children, and about half of people with TSC develop renal cysts over their lifetime. These cysts are small fluid-filled “holes” in the kidney.

A small number of people with TSC develop many cysts that fill up the kidney and affect its function. This is called polycystic kidney disease (PKD). PKD is rare in people with TSC. It arises when a person has a mutation that affects not only the TSC2 gene but also the PKD1 gene, which sits next to it on the same chromosome.
When the cysts are numerous symptoms such as bloody urine, kidney stones and high blood pressure can occur.

Ordinary (so called simple) cysts (fluid-filled bumps) do not need treatment, beyond the usual yearly monitoring of blood pressure and a blood test to check kidney function.

However, in the 5% of people with TSC who have polycystic kidneys (many cysts due to an extra gene defect called PKD1) they may need more frequent monitoring. They should all be referred to a kidney specialist to be assessed for special treatments that are needed in poor kidney function.

Very rarely some people with TSC develop kidney cancer. This can occur at any age. The regular kidney imaging will pick it up. It is slow growing and curable providing it is treated by surgery.

As in AMLs, controlling high blood pressure with medication is very important, because if it is left untreated it can speed up the loss of kidney function.

Information and support

The TSA is here to help and support you.

Living with TSC can raise a number of challenges and uncertainties but much has been learnt about TSC over the last decade and research into the condition has never been so active.

The TSA can help you access the accurate and reliable information that you need, as and when you need it. The TSA can liaise with and signpost you to specialist services and help raise awareness and understanding of TSC among professionals.

Please get in touch with the TSA at www.tuberous-sclerosis.org or by calling 0207 922 7731.
We have a team of TSA Advisers across the UK.

Examples of the ways in which we can help:

- Supporting people attending meetings with professionals, school review meetings, health care meetings, complaints processes and specialist hospital visits
- Helping individuals liaising with social services and health care professionals
- Liaising on your behalf with our professional medical advisers regarding complex TSC queries
- Providing TSC awareness sessions to staff in residential homes, supported living accommodation, schools and to professional staff such as nurses, where appropriate
- Helping to set up local regional support groups
- Holding events and gatherings to exchange information, share experiences and meet others in similar situations

For more information please contact:
support@tuberous-sclerosis.org

Find your regional TSA Adviser here:
www.tuberous-sclerosis.org/tsa_advisers

The Tuberous Sclerosis Association

“To provide hope for today and a cure for tomorrow”
References
A list of TSC scientific references, TSC clinics and TSC professional advisers is available on request.

We value your feedback
Please help us improve the information we offer by sharing your comments on this publication. Please email information@tuberous-sclerosis.org or write to us at the address below.

Supporting the TSA
The TSA provides this information free of charge but if you would like to help cover the cost please visit the fundraising section of our website to make a donation: www.tuberous-sclerosis.org/fundraising.html

Or send a cheque or postal order to:
Tuberous Sclerosis Association, CAN Mezzanine, 32-36 Loman Street, Southwark, London SE1 0EH.

Contact Us
If you would like support or further information about any aspect of TSC, please contact us at:

Tuberous Sclerosis Association, CAN Mezzanine, 32-36 Loman Street, Southwark, London SE1 0EH

By email: admin@tuberous-sclerosis.org
On our website: www.tuberous-sclerosis.org
By telephone: 0207 922 7731

Join the TSC community online:

Facebook: www.facebook.com/thetsauk
Twitter: www.twitter.com/UKTSA
YouTube: www.youtube.com/TSAssociationUK
About the TSA

We’re the only UK charity dedicated to supporting people affected by TSC.

Our aim is to ensure that everyone affected by TSC has the care and support they need.

- **TSCampaign:** Our commitment to raising awareness and driving forward positive change

- **TSCare:** Our commitment to enhancing quality of life for all people affected by TSC

- **TSCure:** Our commitment to research