Malignant Renal Rhabdoid tumours

Information for parents and carers

Grace Kelly Ladybird Trust

This publication is intended to supplement the advice given by your medical team. It was written by Dr Jennifer Kelly, GP and founder of the Grace Kelly Ladybird Trust.

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About this booklet

If you are reading this booklet, the likelihood is that your child, or a child close to you, has recently been diagnosed with a rhabdoid tumour. This leaflet is designed to be a summary of some of the information you may have been already given and it may answer some of your questions. If you have any further worries or queries, please discuss them with the healthcare team looking after your child.

What is a rhabdoid tumour?

A rhabdoid tumour is a rare childhood cancer which may start in the kidneys, but can occur in the brain, spine or other parts of the body. There are 3 main types of rhabdoid tumours grouped together by the locations in which they originate:

1. Malignant renal rhabdoid tumours (MRT) - these occur or originate in the kidney (renal).
2. Extra renal rhabdoid tumours (ERRT) - these occur elsewhere in the body, such as in the liver, lungs, skin or other soft tissues.
3. Atypical teratoid / rhabdoid tumours (AT/RT) - these affect the brain and spinal cord (central nervous system).

Confusingly, the term malignant rhabdoid tumour is sometimes used as a general term to describe extra renal rhabdoid tumours as well. For example, some consultants may refer to a liver (hepatic) rhabdoid tumour as a malignant rhabdoid tumour of the liver or as a hepatic extra renal rhabdoid tumour.

Malignant renal rhabdoid tumours

Malignant renal rhabdoid tumours are tumours that occur in the kidney. They are rare renal tumours that mostly affect infants and very young children.

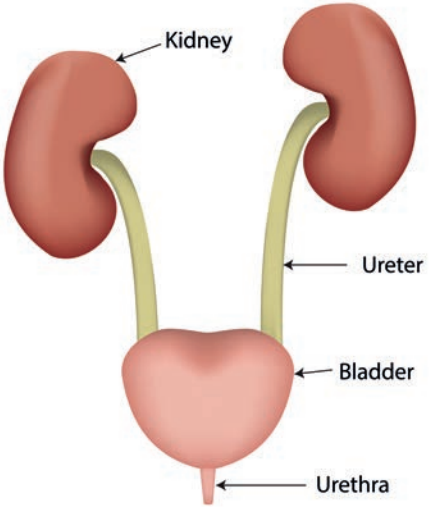
The average age of diagnosis is 15 months old, however renal rhabdoid tumours can occasionally be found in older children. There are currently thought to be about 3 new cases a year in the UK.

Around 1 in 5 children with a malignant renal rhabdoid tumour will also have a brain or spinal cord tumour (multifocal). This may happen in a child with a germ line (genetic) mutation.

What do the kidneys do?

There are two kidneys, one on each side of the spine just below the ribcage. The kidneys clean the blood taking out waste and fluids as urine. The urine produced then passes through the ureter into the bladder and out of the body.

The kidneys also make hormones that help control blood pressure and signal the bone marrow to make red blood cells when needed.



Symptoms at diagnosis

Malignant renal rhabdoid tumours will often present with signs and symptoms of a lump in the abdomen (tummy).

In very young children the presence of pain can be difficult to assess and so symptoms may just be the child appearing more fussy than normal. There also may be visible blood in the urine or high temperatures. In some cases however, none of these symptoms are present.

On examination, the child may have anaemia (low number of red blood cells) or high blood pressure.

What makes it a malignant renal rhabdoid tumour?

Often, on scans, a malignant rhabdoid tumour looks similar to a Wilms Tumour or other renal tumours. It is not until the cells of the tumour are examined under a microscope that a rhabdoid tumour diagnosis can be made.

Malignant rhabdoid tumours have a characteristic genetic change in the cells called a SMARCB1 (or INI1) mutation. It is this change (or mutation) that is responsible for the development of malignant rhabdoid tumours. For more information please see our information booklet *"The genetics of rhabdoid tumours."*

Staging of malignant rhabdoid tumours

Staging is an assessment made by doctors for all patients with cancer to help plan treatment. It categorises the tumour by its location and whether it has spread to any nearby or distant locations in the body. Malignant rhabdoid tumours grow quickly and spread (metastasise) early, therefore many children already have signs of tumour spread at diagnosis. The lungs are a common site for metastases.

Malignant renal rhabdoid tumours are staged as:

Stage I: The tumour is limited to the kidney and has been completely removed. The tumour wall (the capsule) has not been damaged and it has been removed whole. The tumour has not been biopsied prior to it being removed.

Stage II: The tumour may have extended beyond the kidney but has been removed. Some tumour cells may be left behind. There is no spread to the lymph nodes.

Stage III: The main tumour may have spread to local lymph nodes and/or other local structures. The tumour has been biopsied or some has been left behind after surgery.

Stage IV: The tumour has spread to distant lymph nodes or organs. Often this may be in the lungs, liver and brain. Stage IV is the most common stage at presentation.

Stage V: Tumours are present in both kidneys.

What investigations are needed?

The following investigations may be required:

- **Ultrasound scan**
- **CT** (computerised tomography) and/or
- **MRI** (magnetic resonance imaging) scan – usually including the abdomen (tummy), chest and head.
- **Biopsy** of the tumour
- **Blood tests** including kidney tests (GFR)
- **Chest X-ray**
- **ECG** (heart tracing)
- **Echocardiogram** (heart scan)

Treatment of malignant renal rhabdoid tumours

Treatment will normally take place in a specialist care centre that is experienced in treating children with cancer.

Most children will be offered a combination of surgery, chemotherapy and often radiotherapy depending on the tumour location and the age of the child.

Although renal rhabdoid tumours are rare, there are standard treatment guidelines in the UK. If your child has a relapsed tumour, or one that has not responded fully to treatment, you may be offered the opportunity to take part in a clinical trial of a new drug. Your doctor and members of the care team will discuss the options with you in depth.

Surgery

If the child is less than 12 months old, the initial treatment may be to remove the whole kidney with the tumour inside it. In older children, a biopsy (taking a small piece of the tumour) is often done first because other tumour types are more common and may require different treatment.

Once a definite diagnosis of a rhabdoid tumour has been made, the surgeon will want to remove the whole kidney as soon as possible. Removing the whole tumour is always the aim, but this may not be possible straight away.

If the child's tumour is in a difficult location or is too large to remove surgically, chemotherapy is often given first to help shrink the tumour.

Chemotherapy

This is a cancer treatment in which medications are used to kill cancer cells and shrink tumours. Rhabdoid tumours are typically aggressive and can become resistant to chemotherapy quickly. To help reduce this possibility, a combination of chemotherapy drugs are given, often in alternating cycles to help fight the tumour in the most effective possible way.

Radiotherapy

Children over 6 months of age who develop rhabdoid tumours may also receive radiotherapy as part of their treatment.

Supportive care

This is an important part of treatment for childhood rhabdoid tumours. Its role is to help keep the child as comfortable and as free of symptoms as possible. It includes treatment for infections, pain relief and medication to reduce side effects such as sickness.

In many areas as a matter of routine, all children with cancer are referred to the local children's hospice team to help with supportive care (symptom management) whilst on active treatment.

After treatment - follow up

On completing treatment, the frequency of appointments will decrease but there will continue to be regular follow up. This will usually consist of an examination, blood tests and an MRI scan to detect any recurrence.

After treatment, children may face a range of challenges including local effects resulting from the tumour itself as well as the effects of the treatments that they have undergone. The Children's Cancer and Leukaemia Group offer a range of booklets that are very helpful for further information.

Rhabdoid tumours - long term

Statistics tell us the average outcome for children with the same condition. They indicate the proportion of children who would be expected to do well, and the proportion who would be expected not to do so well.

However, it is impossible to predict how each individual child will do. This is why it is important to remember your child is unique and may not follow the expected course of treatment or outcome.

What we know:

- Unfortunately, overall, malignant renal rhabdoid tumours do not have good survival rates.
- Outcomes are poorer if a child has signs of tumour spread at diagnosis.
- Children under 12 months of age who develop rhabdoid tumours are less likely to do as well as older children.
- We cannot predict the exact outcome of each child.

Your child's consultant and medical team will help give you advice and information to make any decisions needed.

If you have any concerns or questions please speak to a member of your child's team.

Grace Kelly Ladybird Trust

The Grace Kelly Ladybird Trust is a UK children's cancer charity that concentrates on funding research and support for children with rare solid tumours. We also work to provide education on the signs and symptoms of childhood cancer and how it may present.

For more information on rhabdoid tumours and a link to our online support group (available for parents of children affected by rhabdoid tumours) please see our website.

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