Atypical Teratoid / 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 an atypical teratoid / rhabdoid tumour (AT/RT). 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 that can start in the brain, spine, kidneys or other parts of the body. There are 3 main types of rhabdoid tumours, grouped together by the locations in which they originate:

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

About Atypical teratoid / rhabdoid tumours

AT/RTs are a very rare type of brain and spinal cord tumour, forming only about 1-2% of brain tumour diagnoses in children. This means for every 100 brain tumours that are diagnosed in children of all ages, only one or two of them would be AT/RTs.

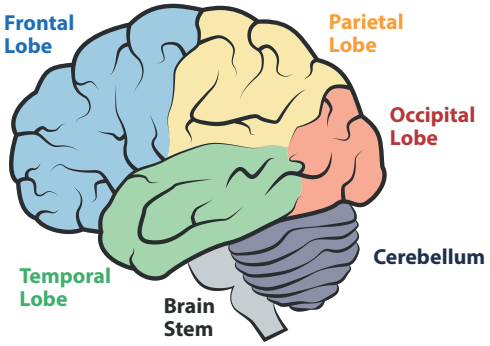
AT/RTs can develop at any age, however, they are most common in very young children. In fact up to 20% of brain tumours occurring in children under the age of 3 will be AT/RTs.

Atypical teratoid / rhabdoid tumours often grow rapidly resulting in the development of symptoms over a relatively short period of time.

What makes it an atypical teratoid / rhabdoid tumour?

Often, on scans, an AT/RT looks similar to other brain and spinal tumours. It is not until the cells of the tumour are examined under a microscope that an AT/RT diagnosis can be made.

All rhabdoid tumours have a characteristic genetic change in the cells called a SMARCB1 (or INI 1) mutation. It is this change that is responsible for the development of rhabdoid tumours. For more information please see our information booklet *"The genetics of rhabdoid tumours."*



Symptoms at diagnosis

Children may present with a range of symptoms including:

- Headaches on waking in the morning (often relieved by vomiting). In babies, this may present as being unusually fussy
- Nausea and vomiting, especially in the morning
- Being unusually sleepy or unable to do skills previously mastered
- An increase in head size in babies
- Blurred or double vision
- In older children, problems with walking, coordination or losing balance

The child's symptoms will vary depending on the location of the tumour.

Around half of these tumours develop in the cerebellum or brain stem (areas towards the lower rear part of the brain). The cerebellum is important in the control of movement, posture and balance.

The brainstem controls the flow of messages between the brain and the rest of the body and also is responsible for controlling basic bodily functions for example breathing, blood pressure and heart rate.

Staging of atypical teratoid / rhabdoid tumours

Staging is an assessment made by doctors for all patients with cancer to help plan treatment. It is used to categorise whether the tumour is in a single place or whether it has spread elsewhere in the brain or the rest of the body.

There is currently no standard staging system for AT/RTs, but tumours are categorised either as:

- **Localised** - occurring in only one location in the brain
- **Metastatic** – has spread to other parts of the brain or spinal cord
- **Multifocal** – rhabdoid tumours develop in more than one location at the same time. This may happen in a child with a germ line (genetic) mutation

Imaging helps to determine whether all or part of the tumour can be removed during surgery (complete or partial resection).

What investigations are needed?

The following investigations may be required:

- **CT** (computerised tomography) and/or
- **MRI** (magnetic resonance imaging), usually of the brain and spine.
- **Lumbar puncture** which removes a small sample of cerebrospinal fluid (CSF) from the spine using a special needle. This is then analysed to assess whether any tumour cells are present in the fluid around the central nervous system. This is usually done under sedation or general anaesthetic.
- **Ultrasound** of the abdomen to check that other organs are healthy and there is no evidence of other tumours.
- **Blood tests**
- **Echocardiogram** (heart scan)
- **Biopsy** or resection (removal) of the tumour
- **Bone scan**

Around 1 in 3 children with an AT/RT will have tumour spread to other parts of the brain or spinal cord at diagnosis. This may make it difficult to remove all of the tumour.

After tissue is removed it is examined and tested under a microscope to tell the difference between AT/RTs and other brain tumours.

Treatment of atypical teratoid / rhabdoid tumours

Treatment will 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 sometimes radiotherapy depending on the tumour location and the age of the child.

Although AT/RTs are rare tumours, 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 doctors and members of the care team will discuss the options with you in depth.

Surgery

This is usually the first step in treating AT/RTs. Depending on the location and size of the AT/RT, the surgeon may only be able to remove part of the tumour.

Chemotherapy

This is a cancer treatment in which medications are used to kill cancer cells and shrink tumours. Atypical teratoid / 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. High dose chemotherapy with a stem cell transplant may also be given.

Radiotherapy

Children who develop atypical teratoid / rhabdoid tumours may have radiotherapy depending on their age at diagnosis. Usually, this is given quite early – within 6–8 weeks of surgery.

Generally, radiotherapy to the brain and spine is avoided in children under 12 months of age and used with caution in children under 3 years of age due to problems it can cause for very young children.

Supportive care

This is an important part of treatment for children with AT/RTs. 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.

Atypical teratoid / 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, atypical teratoid / 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 AT/RTs 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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