Grace Kelly Childhood Cancer Trust

The genetics of rhabdoid tumours

Information for parents and carers

This publication is intended to supplement the advice given by your child's medical team. It was written by Dr Jennifer Kelly, General Practitioner and CEO of the Grace Kelly Childhood Cancer Trust.

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If you would like information on the references used to make this publication, please contact us.

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About this booklet

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

What is a rhabdoid tumour?

Rhabdoid tumours are rare tumours that mainly occur in young children. Rhabdoid tumours include renal rhabdoid tumours, extrarenal rhabdoid tumours, and atypical teratoid rhabdoid tumours (AT/RT).

Most rhabdoid tumours in children occur simply due to chance (sporadic). However, some do occur as part of a hereditary syndrome called rhabdoid tumour predisposition syndrome (RTPS).

If your child has been diagnosed with a rhabdoid tumour, your medical team will discuss with you whether you should be referred to a clinical geneticist who will talk to you about rhabdoid tumours, their genetics and potential risks.

The genetics of rhabdoid tumours

The human body is made up of tiny cells. Each cell contains about 30,000 genes. These carry the information that controls what we look like and how we grow. Genes also have an important role in keeping our bodies healthy. Our genes are stored on chromosomes, a little like books in a library. There are 46 chromosomes (23 pairs) present in each cell of the body.

For us to grow and function, our cells need to multiply. To do this, they make millions of copies of themselves. When this many copies are made, occasionally microscopic errors or alterations can occur. These tiny alterations may be all it takes to stop a cell from functioning properly.

Most damaged cells are identified and destroyed by protective blood cells within the body, but if an abnormal cell is missed and grows rapidly, it is then that cancer can develop.

In rhabdoid tumours, nearly all cases are thought to develop from a copying (translation) error of one gene, the SMARCB1 (INI1) gene. Each cell contains two SMARCB1 genes. Both of these need to be damaged or altered to result in the development of a rhabdoid tumour in any location.

It is very rare for this to occur unless a child has been born with one of their SMARCB1 genes already altered or damaged (RTPS). This is a germline mutation.

Occasionally both SMARCB1 genes can be damaged even though a child was not born with a germline mutation. This is called a sporadic tumour.

About rhabdoid tumour predisposition syndrome

Rhabdoid tumour predisposition (RTPS) is found in around a third of children who are diagnosed with a rhabdoid tumour. This means that for every 10 children diagnosed with a rhabdoid tumour, 3 of them will have RTPS.

Typically, children with RTPS are likely to be diagnosed at an earlier age than children with sporadic (no genetic cause) tumours. They also have an increased chance of developing multiple tumours and generally do not have as good outcomes as children without RTPS.

What is rhabdoid tumour predisposition syndrome?

RTPS is almost always caused by an alteration (or mutation) that has occurred within the SMARCB1 (INI1 gene). Most individuals have two healthy copies of the SMARCB1 gene, located on chromosome 22.

The SMARCB1 gene controls the production of proteins which act as "tumour suppressors". These help keep cells healthy and stop them from growing and dividing too quickly.

In individuals with RTPS, one copy of the SMARCB1 gene is altered in every single cell of the body leaving only one healthy copy (instead of two). If the healthy copy of the SMARCB1 gene then gets damaged by chance, rapid cell growth can occur resulting in a rhabdoid tumour.

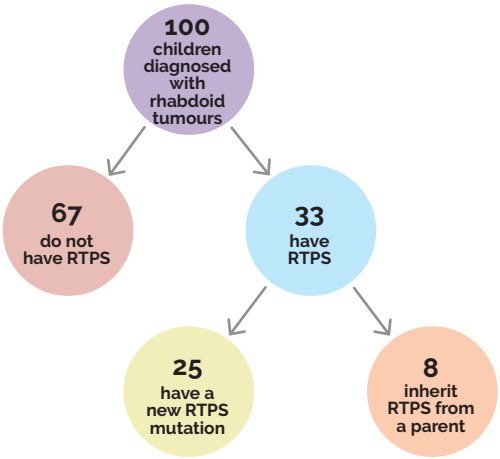
Very rarely, a mutation can occur on chromosome 19, known as a SMARCA4 mutation. This also leads to the development of RTPS in a similar way.

Rhabdoid tumours develop due to chance and are not caused by anything that a child has been exposed to. They cannot be avoided or prevented.

My child has rhabdoid tumour predisposition syndrome. Where has it come from?

Some children with RTPS have inherited an altered copy of the SMARCB1 gene from a parent who carries the same genetic mutation (but may not know about it). Usually, however, the RTPS results from the development of a 'new mutation' in either the egg or sperm before a child has been conceived or in the cells of the developing embryo.

If this is the case, your child would be the first person in the family to carry this genetic change, and other family members should not be at increased risk of developing rhabdoid tumours.



All individuals with RTPS have a 50% chance of passing the condition on to their children whether they inherited it themselves or whether it was a new mutation.

Children who inherit the altered copy of the gene will also have RTPS and be at increased risk of developing rhabdoid tumours. Children who do not inherit the altered gene should not be at increased risk.

Testing for rhabdoid tumour predisposition syndrome

The simplest way to test for RTPS is from a sample of blood. Specific cells in the blood are then identified and the genetic information is analysed. Both copies of the SMARCB1 gene are checked for mutations.

Any mutations found are then compared with the mutation found in the tumour tissue itself. If the blood sample does not contain the same genetic mutation as the tumour sample, the child does not have RTPS.

If an individual is found to have RTPS, blood samples from other family members can also be tested. If neither parent carries the mutation, it means that the RTPS was not inherited and must have occurred as a new mutation. This means that no other family members should be at risk of having RTPS.

Around a quarter of cases of RTPS (25%) are thought to be inherited. The remaining three quarters of cases (75%) are thought to be sporadic (new mutation).

It is important to consider whether or not you would want to find out this information. Discussing the options with a geneticist is advised.

What does having rhabdoid tumour predisposition syndrome mean?

Children with RTPS are at increased risk of developing rhabdoid tumours. This risk is at its highest under the age of 5 years. Not all children with RTPS will definitely develop a rhabdoid tumour, but they are at a significantly higher risk.

Older children and adults with RTPS are also at increased risk of developing schwannomas (tumours of nerve tissue).

It is not known why some individuals with RTPS develop rhabdoid tumours and others develop schwannomas, but it is likely to be a result of a combination of factors.

Do individuals with RTPS need monitoring if they are otherwise well?

Recent research has suggested that individuals with RTPS should receive regular monitoring including a whole-body MRI at diagnosis.

Additional monitoring may include:

- For infants (less than 12 months old), ultrasounds of their tummy and neck approximately every 2 to 3 months.
- For children, frequent MRI scans of the brain and spine or alternatively whole-body MRI scans.

This monitoring may differ depending on your child's needs and so will be decided with your child's medical team.

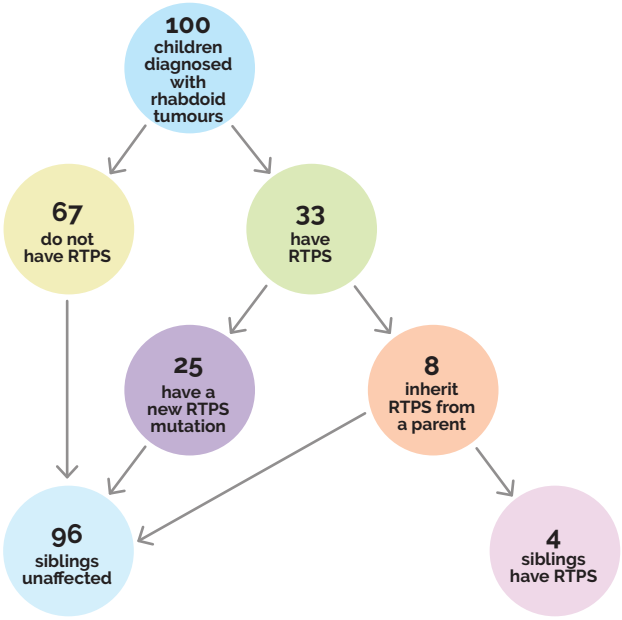
My child has a rhabdoid tumour. Will my other children develop them too?

It is unlikely that any siblings your child has will develop rhabdoid tumours.

In some situations, however, they may be at increased risk if your child with the rhabdoid tumour has RTPS. If your child with the rhabdoid tumour does not have RTPS, your other child(ren) should not be at increased risk. Your team will give you further advice on this.

As illustrated below, if you take 100 children with rhabdoid tumours, 33 will have rhabdoid tumour predisposition syndrome, and just 4 will have a sibling with RTPS.

The possibility of children with rhabdoid tumours having a sibling with rhabdoid tumour predisposition syndrome (RTPS)*



*Numbers are approximate to illustrate proportions

Is there a test for rhabdoid tumour predisposition syndrome during pregnancy?

If you have a child with a rhabdoid tumour but your child does not have RTPS, the chance of you having another child with a rhabdoid tumour is extremely unlikely. Testing in future pregnancies is not usually recommended in these circumstances.

If you have a child with a rhabdoid tumour and this child does have RTPS, but it was not inherited from either parent, the chance of having another baby affected by a rhabdoid tumour is small. Testing can be offered in future pregnancies if you are concerned.

If a parent carries the SMARCB1 gene change themselves (a parent has RTPS), there would be an increased risk of having another baby with a rhabdoid tumour. In this situation, couples can consider IVF-based techniques to conceive a baby or choose testing in pregnancy.

Please discuss this with your geneticist if you would like more information.

Support for you

As a parent or carer, this is a very difficult time for you. Many parents talk about going into survival mode when their child has been diagnosed with cancer, but it is important to take time to look after yourself when you can.

Talking to loved ones or professionals and by asking for practical help when you need it, can really make a difference. If you do not know where to turn, please speak to your child's medical care team or your General Practitioner for more support and advice.

For more information on booklets we produce, for more information on rhabdoid tumours and for a link to our online rhabdoid parents support group, please see **www.gkcct.org**

Grace Kelly Childhood Cancer Trust

The Grace Kelly Childhood Cancer Trust is a UK-based children's cancer charity that concentrates on funding research and providing support to families. We teach the signs and symptoms of childhood cancer and are working towards earlier diagnosis of children through the education of parents, carers and clinicians.

Our medical publications are written by medics and are extensively peer-reviewed. Bridging the gap between parents and medics.

For more information on our work and the publications we have produced, please see our website.

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