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# Juvenile Myelomonocytic Leukaemia (JMML)

**A Guide for  
Parents**

**Leukaemia Care**  
YOUR Blood Cancer Charity

# Introduction

**Being told your child has juvenile myelomonocytic leukaemia (JMML) can be a shock, particularly when you may never have heard of it. If you have questions about JMML – what causes it, who gets it, how it affects your body, what symptoms to expect and likely treatments – this booklet covers the basics for you.**

You will also find useful advice about how to get the best from your child's haematologist, plus practical advice on how to help important people in your child's life understand such a rare condition. For more information, talk to your child's haematologist or clinical nurse specialist.

Booklet originally written by Isabelle Leach, our Patient Information Writer and peer reviewed by Professor Mary Frances McMullin. We are also grateful to Jazzmyn Saunders, whose son has JMML, for her contribution as a reviewer.

If you would like any information on the sources used for this booklet, please email [communications@leukaemiacare.org.uk](mailto:communications@leukaemiacare.org.uk) for a list of references.

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# About Leukaemia Care

**Leukaemia Care is a national charity dedicated to ensuring that people affected by blood cancer have access to the right information, advice and support.**

## Our services

### Helpline

Our helpline is available 9.00am - 10.00pm on weekdays and 9.30am - 12.30pm on Saturdays. If you need someone to talk to, call **08088 010 444**

### Nurse service

We have two trained nurses on hand to answer your questions and offer advice and support, whether it be through emailing **nurse@leukaemicare.org.uk**, over the phone on **08088 010 444** or via LiveChat.

### Patient Information Booklets

We have a number of patient information booklets like this available to anyone who has been affected by a blood cancer. A full list of titles – both disease specific and general information titles – can be

found on our website at **www.leukaemicare.org.uk/support-and-information/help-and-resources/information-booklets/**

### Support Groups

Our nationwide support groups are a chance to meet and talk to other people who are going through a similar experience. For more information about a support group local to your area, go to **www.leukaemicare.org.uk/support-and-information/support-for-you/find-a-support-group/**

### Buddy Support

We offer one-to-one phone support with volunteers who have had blood cancer themselves or been affected by it in some way. You can speak to someone who knows what you are going through. For more information on how to get a buddy call

**08088 010 444** or email  
**support@leukaemicare.org.uk**

### Online Forum

Our online forum, **www.healthunlocked.com/leukaemia-care**, is a place for people to ask questions anonymously or to join in the discussion with other people in a similar situation.

### Patient and carer conferences

Our nationwide conferences provide an opportunity to ask questions and listen to patient speakers and medical professionals who can provide valuable information and support.

### Website

You can access up-to-date information on our website, **www.leukaemicare.org.uk**, as well as speak to one of our care advisers on our online support

service, LiveChat (9am-5pm weekdays).

### Campaigning and Advocacy

Leukaemia Care is involved in campaigning for patient well-being, NHS funding and drug and treatment availability. If you would like an update on any of the work we are currently doing or want to know how to get involved, email **advocacy@leukaemicare.org.uk**

### Patient magazine

Our quarterly magazine includes inspirational patient and carer stories as well as informative articles by medical professionals. To subscribe go to **www.leukaemicare.org.uk/communication-preferences/**

# What is JMML?

**Juvenile myelomonocytic leukaemia (JMML) is an excessive production of the monocyte white blood cells in the bone marrow, which infiltrate other organs including the spleen, liver, lung, and gastrointestinal tract.**

The term JMML includes all diagnoses previously known as:

- Juvenile chronic myeloid leukaemia (JCML)
- Chronic myelomonocytic leukaemia of infancy (CMMoI)
- Infantile monosomy 7 syndrome

The diagnostic criteria of JMML are included in the 2016 World Health Organisation (WHO) classification of myeloid neoplasms (bone marrow cancers) and acute leukaemia (go to the section on **diagnosis** for these diagnostic criteria).

JMML represents 2% to 3% of all childhood leukaemias, with an incidence between 0.6 to 1.2 per million per year for children aged 0 to 14 years. Around 75% of patients present below three years of age.

Monocytes normally make up 5%

to 10% of the cells in human blood. Together with the neutrophil white blood cells, monocytes enter the body's tissues to attack invading organisms and help combat infections. Around 40% of JMML cases present before one year of age and boys are affected more often than girls by a ratio of 2.5 to 1.

In JMML, an excess of stem cells in the bone marrow become monocytes, leading to increased numbers of monocytes in the blood, bone marrow and other organs. Some of these stem cells develop as immature monocytes called blasts. This surplus of monocytes can interfere with the production of the other healthy blood cells such as the red blood cells, platelets and the other white blood cells in the bone marrow.

The majority of children with JMML receive an allogeneic stem cell transplant, which

is potentially a cure for many youngsters.

## What causes JMML?

Around 90% of children with JMML have particular genetic mutations. In the remaining 10%, there is no identifiable gene mutation. The genes where mutations have been identified include the following:

- **K-RAS (Kirsten Rat Sarcoma), N-RAS (Neuroblastoma Rat Sarcoma):** These genes act in the signalling of cells to control cell growth and cell death. Approximately 20% - 25% of patients with JMML have a mutation in a gene of the RAS gene family.
- **CBL (Casitas B-lineage Lymphoma):** The CBL gene is a protein that functions as a tumour suppressor, and is also known to prevent normal immune responses turning into autoimmune diseases. Children with CBL mutations have a high rate of their JMML resolving spontaneously. They also have diminished growth, developmental delays, undescended testes and

vasculitis (inflammation of the blood vessels). About 10% - 15% of patients with JMML have a mutation of the CBL gene.

- **NF1 (Neurofibromatosis type 1):** This gene functions as a tumour suppressor, and in some patients with a mutation in this gene, the condition neurofibromatosis type 1 is present. NF1 is characterised by multiple café-au-lait (milky-coffee) coloured spots, benign or malignant tumours, cognitive and behavioural problems, and skin fold freckling (freckles in areas not exposed to the sun). Around 15% - 20% of patients with JMML have a mutation in the NF1 gene.
- **PTPN 11 (Protein Tyrosine Phosphatase, Non-receptor type 11):** Mutation in this gene leads to the Noonan syndrome, which is characterised by unusual facial features, short stature, skeletal anomalies, heart defects, learning difficulties, and bleeding disorders. Up to 35% of patients with JMML have a mutation in the PTPN 11 gene. While in most cases JMML caused by a mutation is known to be

# What is JMML? (cont.)

aggressive, in JMML occurring in patients with Noonan syndrome, the cancer is often benign and transitory.

- **Chromosome 7 deletion:**  
Around 25% of children with JMML have been identified as having a deletion of chromosome 7.
- **Additional mutations:**  
Research has shown that patients who have additional mutations (also called secondary mutations) such as in the SETBP1 (SET binding protein 1) gene and the JAK3 (Janus Kinase 3) gene are less likely to achieve a cure compared with those with only one mutation.

# Symptoms and diagnosis of JMML

JMML generally progresses slowly, so there may be few symptoms at the onset. The most common symptoms of JMML are listed below; however, children with JMML can show any combination of these symptoms:

- Pale appearance
- General fatigue or weakness
- Decrease in appetite and/or weight loss
- Irritability
- Fever
- Rash
- Recurrent infections
- Bruising easily or bleeding
- Rash
- Developmental delays
- Enlarged liver, spleen or lymph nodes
- Abdominal pain, bone and joint pain (due to overcrowding with monocytes)

Symptoms can appear over weeks or months. Patients who have neurofibromatosis type 1

or Noonan's syndrome will also display the symptoms specific to those conditions as described previously.

## Diagnosis of JMML

To make a definite diagnosis of JMML, the criteria set out in the 2016 WHO classification of JMML of myeloid neoplasms and acute leukaemia must be met. They are as follows:

### JMML diagnostic criteria - 2016 WHO classification

#### 1. Clinical and haematologic features (All four features are mandatory)

- Peripheral blood monocyte count  $\geq 1 \times 10^9/L$
- Percentage of blast cells in peripheral blood and bone marrow  $< 20\%$
- Splenomegaly (enlarged spleen)
- No Philadelphia chromosome, also called re-arrangement of BCR-ABL1 (Breakpoint Cluster Region-Abelson Murine Leukaemia Viral proto-oncogene 1)

#### 2. Genetic studies (One finding

# Symptoms and diagnosis of JMML (cont.)

## is sufficient)

- Somatic mutation in PTPN11, K-RAS or N-RAS (Noonan syndrome must be excluded)
- Clinical diagnosis of NF1 or NF1 mutation
- CBL mutation and loss of heterozygosity (genetic variability) of CBL

### **3. For patients without genetic features, besides the clinical and haematologic features listed under 1, the following criteria must be fulfilled:**

- Monosomy 7 or any other chromosomal abnormality, or at least two of the following criteria:
- Haemoglobin F (foetal haemoglobin) increased for patient's age
- Precursors of bone marrow cells or red blood cells seen on peripheral blood smear. Precursor cells are a type of partially differentiated cell which has the capacity to differentiate into only one cell type.
- Granulocyte-macrophage-

colony-stimulating factor hypersensitivity in colony assay present

- Hyperphosphorylation of STAT5 (Signal Transducer and Activator of Transcription 5)

Prior to reaching a diagnosis of JMML as above, other diagnoses are generally excluded, particularly if the patient is older than the average two years at diagnosis. JMML is diagnosed by the use of blood tests, bone marrow aspiration and biopsy, including analysis of chromosomes and their abnormalities.

## Full blood count

Also called a complete blood count, this measures the numbers of red blood cells, white blood cells, platelets as well as the level of haemoglobin and the haematocrit, which is the proportion of red blood cells to plasma (fluid component in the blood).

- A number of children with JMML have elevated Haemoglobin F levels for their age.
- White blood cells in patients with JMML may be abnormal, but a bone marrow examination is needed for confirmation.

## Bone marrow examination

A bone marrow biopsy is the collection of a sample of bone marrow from the hip bone, generally under local anaesthesia, and its examination under the microscope to determine the number and type of cells present and the level of haematopoiesis (process by which blood cells are formed). A percentage of blast cells <20% is required as part of the diagnosis of JMML.

## Additional testing

Other tests may be conducted to assess the patient's general health and performance of the vital organs. These include:

- **Further blood tests:** Blood chemistries, and liver and kidney function tests.
- **Imaging tests:** X-rays, computer tomography (CT) scans, and magnetic resonance imaging (MRI) scans.
- **Genetic testing, with particular reference to the characteristic RAS, PTPN1, NF1, CBL or BCR/ABL1:** This helps differentiate JMML from other types of leukaemia. This also enables treatments to be tailored to your

specific condition.

- **Spinal tap/lumbar puncture:** A needle is inserted into the spinal canal to access the area around the spinal cord to measure and draw off a small amount of cerebral spinal fluid to test for suspected infections.
- **Human leukocyte antigen (HLA) typing:** Also called tissue typing, this process is used to find a suitable donor, should a stem cell transplant be needed. The proteins on the surface of the patient's blood cells are matched with those of a potential donor. The greater the number of HLA markers that are shared by the patient and donor, the better the chance of a successful transplant.
- **GM-CSF (Granulocyte-macrophage colony-stimulating factor) hypersensitivity assay:** GM-CSF is a growth factor which stimulates the growth of living cells. Increased amounts of GM-CSF are added to samples of blood or bone marrow in which healthy cells will not grow, but JMML cells will grow. However, this test takes weeks to complete, is not standardised and is not widely available.

# Treatment options for JMML

Due to the rarity of JMML and the recent findings of chromosomal mutations linked to it, a standard treatment consensus for JMML is still awaited.

At present, the only successful treatment option for the cure of JMML is an allogeneic stem cell transplant (ASCT). Chemotherapy may alleviate symptoms but cannot offer a cure. Splenectomy before transplantation has not been shown to be of benefit.

## Stem cell transplantation

The only effective treatment for JMML currently is an ASCT, which achieves a cure in approximately 50% of patients. Without an ASCT, median survival time for patients with JMML is around one year.

An ASCT is indicated for the majority of children with JMML, particularly for those with NF1 and somatic PTPN11 mutations, and most of those with somatic K-RAS mutations and somatic N-RAS mutation. A certain proportion of patients with somatic N-RAS mutation or CBL mutation have spontaneous regression; therefore, a 'watch and wait'

strategy is more appropriate for these patients.

A better outcome with an ASCT is often seen for patients of a younger age. However, an ASCT for these children carries the risk of severe toxicity, during the procedure and later in life.

## Conditioning regimen

Prior to children patients receiving the infusion of donor blood cells as part of their ASCT, they normally receive a conditioning regimen. This usually consists of high-dose chemotherapy to eliminate the cancer cells and prevent the immune system rejecting the new stem cells. A period of two to four weeks is generally needed for the stem cells to multiply and make new blood cells, a process which is called engraftment.

Patients are generally conditioned for the ASCT with a regimen of busulfan, cyclophosphamide and melphalan. Total body irradiation is sometimes used as part of the conditioning regimen; however, this is controversial given the possible side effects later in life, such as short stature, learning

difficulties, secondary cancers, and sterility. The Children's Oncology Group (COG) JMML study includes total body irradiation and cyclophosphamide as there is a fear that chemotherapy on its own may not be sufficient to eliminate inactive JMML blast cells.

### Graft-versus-host disease

Graft-versus-host disease is a serious problem that occurs with ASCTs. It happens when the graft (donated marrow or stem cells) reacts against the host (patient receiving the stem cells). The T-cells in the donated stem cells attack and destroy the host's cells as they see them as foreign bodies. Symptoms of graft-versus-host disease include skin rashes, diarrhoea and liver damage. Graft-versus-host disease can be very mild and short-lived (acute form) or it can be severe and even life threatening, lasting for years (chronic form).

Graft-versus-host disease has been shown to have an important role in the treatment of JMML. Acute or chronic graft-versus-host disease is linked to a lower relapse rate in patients with

JMML. Children who receive fewer immunosuppressant drugs in the conditioning regimen have lower relapse rates.

Small studies have reported that not using melphalan and/or substituting cyclophosphamide with fludarabine in the conditioning regimens may decrease acute graft-versus-host disease without affecting overall survival.

### Relapse and second ASCT

A relapse represents the main failure of an ASCT for a child with JMML, with the rate being as high as 50%. ASCTs using HLA matched family donors, HLA-matched unrelated donors and HLA-matched unrelated umbilical cord blood donors have generated similar relapse rates, which means that the lack of an HLA matched donor does not prevent good ASCT outcomes. However, despite recent reductions in ASCTs-related deaths, the deaths are still higher with HLA matched unrelated donors, mostly due to infection.

Factors which predict an increased risk of relapse are an

# Treating options for JMML (cont.)

age of over four years and having a bone marrow blast percentage greater than 20%. A second ASCT may achieve a cure, particularly when combined with decreased immunosuppression, as this produces a stronger graft-versus-leukaemia effect. Second ASCTs were successfully carried out in 15 patients from the European Working Group/European Bone Marrow Transplant Group (EWOG/EBMT) trial, using the original donor as often as possible, and total body irradiation being the most common conditioning regimen. However, the intentional reduction in the cyclosporine conditioning regimen to prevent relapse did lead to an expected high rate of graft-versus-host disease.

In a series of five case reports where the conditioning regimen was high-dose cytarabine plus mitoxantrone, good results were reported following repeat cell infusion from the original donor.

These findings suggest that relapse does not inevitably mean a poor prognosis and that a second ASCT is a valid option

for patients in good physical condition. However, whether or not the same donor would be used, if this was feasible, would be up to the discretion of your child's medical team and what they feel would provide the best outcome for your child.

## Chemotherapy

Without treatment, around one third of patients with JMML will progress rapidly leading to early death. However, there have been reports of some patients remaining in a stable condition despite not receiving treatment for up to 12 years. Nevertheless, without an ASCT, survival time of children with JMML is 10 to 12 months.

Two JMML treatment protocols are commonly used, although they are not internationally accepted as yet:

- 1. The European Working Group of Myelodysplastic Syndromes in Childhood (EWOG-MDS) study protocol:** An ASCT after a conditioning regimen of busulphan, cyclophosphamide and melphalan.

## 2. The North American Children's Oncology Group (COG) study

**protocol:** An ASCT after conditioning with combination of busulfan and fludarabine.

- The COG conducted a clinical trial comparing their study treatment protocol with that of EWOG-MDS study treatment to establish if their protocol is associated with less transplantation-related mortality but comparable disease-free survival. The trial ([ClinicalTrials.gov: NCT01824693](https://clinicaltrials.gov/ct2/show/study/NCT01824693)) was closed prematurely after patients treated with busulfan and fludarabine were noted to have increased rates of disease progression.

Busulfan, cyclophosphamide, and melphalan are therefore currently recommended as a conditioning regimen for all patients undergoing an ASCT until an appropriate reduced-toxicity regimen can be identified.

Long term remission of JMML with chemotherapy treatment alone has not been achieved. Nevertheless, chemotherapy can

improve the symptoms of JMML in patients who do not suffer from an aggressive form of the disease. Patients with JMML may be given 6 mercaptopurine or low-dose intravenous cytarabine to control their symptoms; however, responses are usually temporary. In addition, chemotherapy treatment is given as part of the conditioning regimen prior to an ASCT.

## Splenectomy

Splenectomy prior to an ASCT for the purpose of avoiding a relapse has been tried in several patients with JMML. The reasoning for a splenectomy is that the numerous white blood cells in the enlarged spleen will prevent dormant JMML cells from being eliminated by irradiation therapy or chemotherapy, thus leading to relapse. However, results of the EWOG-MDS/EBMT study showed that the size of the spleen at the time of the ASCT, and performing a splenectomy prior to the ASCT, did not influence the risk of relapse or the probability of survival.

# Treating options for JMML (cont.)

## New treatments

Research into alternative treatments for JMML is focussed on targeted therapies (drugs that specifically interrupt the ability of the leukaemia to grow in the body) and immunotherapies (treatment that uses the body's own immune system to fight the cancer). These include:

- **Azacitidine:** This anti-cancer drug works by 'switching on' genes that prevent the cancer cells growing and dividing. In February 2011, it was approved by the National Institute for Health and Care Excellence (NICE) in the UK for the treatment of myelodysplastic syndromes, chronic myelomonocytic leukaemia and acute myeloid leukaemia in adults. It is also being investigated as a treatment for JMML.
- **13-cis retinoic acid:** This retinoid drug (related to Vitamin A) is known to inhibit the growth of JMML cells in the laboratory. The COG JMML study has included 13-cis-retinoic acid in its treatment protocol, though its therapeutic value for JMML

remains controversial.

- **Tipifarnib:** This is a farnesyl transferase inhibitor that works by blocking the enzymes necessary for cancer cell growth. In a recent study, administration of tipifarnib followed by an ASCT in patients with newly diagnosed JMML was found to be safe and yielded a 51% initial response rate as a single agent, but failed to reduce relapse rates or improve long-term overall survival.
- **Trametinib:** This is a MEK (mitogen-activated protein kinase kinase enzyme) inhibitor which is being investigated in a COG-sponsored trial, for children with relapsed or refractory JMML.

Other agents that are being investigated for use in JMML include RAS mimetics, SHP-2 (Src homology phosphotyrosine phosphatase 2) inhibitors, anti-GM-CSF antibodies, and chimeric antigen receptor (CAR) T-cell therapy, which involves genetically engineering the patient's T-cells to target and kill leukaemia cells.

## Prognosis

For children with JMML, the only possibility of a cure is an ASCT. Relapse will occur in up to 50% of patients following the ASCT.

For patients with JMML, factors which have been linked to a poor prognosis (i.e. high likelihood of relapse) include:

- Age >2 years old at diagnosis
- SETBP1 and JAK3 mutations, particularly SETBP1
- Low platelet count <33 x 10<sup>9</sup>/L, (normal range: 150-400 x 10<sup>9</sup>/L)
- High haemoglobin foetal (F) levels ≥ 10%, (normal range: 0.3 to 4.4 %)

As with most cancers, prognosis can vary greatly. Early presentation, proactive medical attention on presentation and aggressive therapy are important for the best prognosis. New treatments are continually being discovered to improve treatment and decrease side effects of an ASCT and conditioning regimens.

# Living with JMML

**After your child's diagnosis of JMML, you may find that it affects you emotionally. This chapter will talk about how receiving this news can impact you and your child.**

## Emotional impact of JMML

Being told your child has cancer can be very upsetting. Seeing your child with some of the symptoms of JMML can be hard to cope with and, because of this, you may need emotional, as well as practical, support. Your child's diagnosis with a rare disease can affect you emotionally at any point of your child's journey. It is likely that you will experience a range of complex thoughts and emotions, some of which may feel strange or unfamiliar to you. It is important to know that these feelings are all valid and a normal response to your situation.

## Looking after your child and your family

Following a diagnosis of JMML you may want to make changes to your child's routine to ensure the best health of your child after the diagnosis and during treatment. Don't try to change too much at

once. Adopting a healthy routine for your child is about making small, manageable changes.

A healthy lifestyle includes a well-balanced diet and remaining active. With some of the side effects your child may be experiencing, the idea of going out to play and being active may be the last thing your child wants to do, but it is important to try and stay as active as possible to make your child feel better and distract them from some of the symptoms or side effects.

One of the most commonly experienced side effects of the treatment of JMML is fatigue. This is not normal tiredness and does not improve with sleep.

Some general tips on how to deal with your child's fatigue include:

- Have a regular lifestyle – going to bed and waking up at approximately the same time every day
- Take part in regular, gentle play

to maintain your child's fitness level as much as possible

- Build rest periods in your child's day to preserve their energy for what is important
- Before going to bed, avoid stimulating activities such as television, or using laptops, tablets or mobile phones, if applicable
- Keep your child's bedroom quiet and at a comfortable temperature
- Talk to your child about their worries
- Discuss your child's symptoms with your doctor or nurse

## Practical support Work and finances

If your child is diagnosed with JMML, it can sometimes lead to difficulties relating to your work life. You may want to reduce working hours, but it can also mean that you have to stop work altogether. You may need to make an arrangement with your employer for times when you can accompany your child into hospital.

It is often worth taking time to explain your child's JMML to your employer, as it is likely they will never have heard of the disease. Your child's consultant or your GP can arrange letters to confirm your child's diagnosis to help you explain your situation to your employer.

Macmillan has published a booklet about financial support following a child's diagnosis of cancer. They can also give you personal advice over the phone via their helpline on 0808 808 00 00 and you can discuss which benefits you are eligible for. Some Macmillan centres can arrange face-to-face meetings with a benefits advisor. They can also provide financial assistance in the form of grants – ask your nurse in the hospital how to apply.

If you would like more information about some of the things you may be entitled to, you can speak to our Advocacy Officer by emailing [advocacy@leukaemicare.org.uk](mailto:advocacy@leukaemicare.org.uk) or by calling 08088 010 444.

# Talking about JMML

## Talking to the haematologist

JMML is a rare condition. It is important for you to develop a good working relationship with the haematologist, so your child is given the best treatment possible.

The following gives advice on working well with your child's haematologist:

- If it is an initial consultation, take along a list of your child's current medications and doses, and a list of any allergies your child may have.
- If your child has a complicated medical history, take a list of diagnoses, previous procedures and/or complications.
- Make a list of questions to take to the appointment. This will help the discussion with your child's haematologist.
- It can be useful to repeat back what you have heard so that you can be sure that you fully understand what the next steps are for your child.
- Note information down to help

you remember what was said.

- Be open when you discuss your child's symptoms and how you and your child are coping. Good patient-doctor communication tends to improve outcomes for patients.

Other tips:

- Bring someone else along to your child's appointment – they can provide support, ask questions and take notes if you are focussing on your child in the appointment.
- Do not be afraid to ask for a second opinion – most haematologists are happy for you to ask.

## You need to tell your haematologist if...

Your child is having any medical treatment or taking any products such as prescribed medicines, over the counter treatments or vitamins. It is important to understand that treatments, including complementary therapies, which are perfectly safe for most children, may not be safe if your child is being treated for

## JMML

Remember, if you choose to start your child on any form of complementary treatment outside of their medical treatment, discuss this with their haematology consultant or clinical nurse specialist, prior to beginning it. It is important to understand the difference between complementary therapies, used alongside standard treatment, and alternative therapies, used instead of standard treatment. There is no evidence that any form of alternative therapy can treat JMML.

## Talking to other people

- Telling people that your child has a rare condition like JMML can be hard to explain. You might find it useful to let your close family and friends, as well as your employer, know about your child's health condition. It might be easier to provide people with basic information and give them information leaflets or a booklet like this one about JMML if they want to know more in-depth details.

- It is probably best to focus conversations on the symptoms that your child is experiencing, how the condition affects them and how they feel about it. Often people misunderstand and, unfortunately, it will mostly fall to you to educate them as best as you can. Where possible, it's advisable to let people know what you find helpful and unhelpful, in terms of what others say and do. Often people make assumptions and do what they think helps. For example, saying your child looks well, recounting stories of others they know with a similar diagnosis, encouraging you to look ahead and stay positive is not always what people really want to hear. In many ways, the more you communicate with them, the better.

These points may help you:

- Explain that your child has a condition that means their bone marrow does not function properly, and that this affects the number of blood cells it produces
- Explain your child's symptoms (maybe they are tired, or have a

lot of pain)

- Explain what you and your child need (maybe more help day-to-day, or someone to talk to)

You could also consider the following when telling people about your child's diagnosis:

- **Find out more** - Try to find out as much as you can about your child's condition, from reliable internet sources, charitable organisations or your consultant haematologist. The more you know, the more you can share.
- **Have a print-out to hand** - It may help to have some information to hand to share with family and friends. This will take the pressure off you having to remember everything they may want to know.
- **Explain yours and your child's needs** - Try and be clear about what your needs may be. Perhaps you need help with the weekly food shop, help with cooking dinner, or someone to drive you to and from your child's appointments. You may find that friends and family

are pleased that they can do something to help you.

- **Be open about how you feel** - Don't be afraid of opening up about how you feel, as people who care will want to help you as best they can. Talk as and when you feel comfortable, so those around you will know when you need them most.

If you're struggling to come to terms with your diagnosis and prognosis, you can speak to us on our helpline. Call us on **08088 010 444**

# Glossary

## Allogeneic stem cell transplant (ASCT)

Stem cell transplant of cells from a matching donor.

## Amino acids

Organic molecules which are the building blocks for making proteins.

## Antibody

Blood protein produced by the B-cell lymphocytes in response to, and fighting, a specific antigen, such as a bacteria, virus, or foreign substance in the blood.

## Autologous stem cell transplant

Stem cell transplant of cells derived from part of the same individual.

## Blasts

Patients with leukaemia have a high number of abnormal white blood cells. These white blood cells are not fully developed and are called blasts.

## Café-au-lait spots

Milky coffee (in French) coloured spots typically seen in patients

with neurofibromatosis type 1.

## Chemotherapy

Drugs that work in different ways to stop the growth of cancer cells, either by killing the cells or by stopping them from dividing.

## Chromosomes

Thread-like structures which carry the genes, and are located in the nuclei of every cell in the body. There are 46 chromosomes (23 pairs) in humans.

## ClinicalTrials.gov

ClinicalTrials.gov is a database of trials and includes details of 276,190 research studies in all 50 states and in 204 countries.

## Conditioning regimen

Chemotherapy or total body irradiation to eliminate the cancer cells and prevent the immune system rejecting the new stem cells prior to an allogeneic stem cell transplant.

## DNA (deoxyribonucleic acid)

Thread-like chain of amino acids found in the nucleus of each cell in the body which carries genetic instructions used in the growth,

# Glossary (cont.)

development and functioning of the individual.

## Engraftment

Process by which stem cells from a donor multiply and make new blood cells.

## Fatigue

Tiredness and weakness.

## Graft-versus-host disease

Serious complication that occurs with allogeneic stem cell transplants. It happens when the graft (donated marrow or stem cells) reacts against the host (patient receiving the stem cells).

## Granulocyte-macrophage colony-stimulating factor (GM-CSF)

Growth factor required to stimulate the growth of living cells.

## Haematopoiesis

Process by which blood cells are formed.

## Haemoglobin F

Foetal haemoglobin. Normal range: 0.3% to 4.4% of haemoglobin.

## Human leukocyte antigen (HLA) typing

Also called tissue typing, this process is used to find a suitable donor for stem cell transplantation. The proteins on the surface of the patient's blood cells are matched with those of a potential donor. The greater the number of HLA markers that are shared by the patient and donor, the better the chance of a successful transplant.

## Immunotherapy

Treatment that uses the body's own immune system to fight the cancer.

## Irradiation

Particles or rays falling on to a surface (e.g., radiation wave on the surface of the skin).

## Juvenile myelomonocytic leukaemia (JMML)

Excessive production of monocyte white blood cells in the bone marrow, which infiltrate other organs such as the spleen, liver, lung, and gastrointestinal tract.

## Lymph nodes

Components of the lymphatic system (part of the body's

immune system) that contain lymphocytes which produce antibodies and macrophages to digest dead cells. Lymph nodes are swollen with cell fragments in the event of infection or cancer. They are located mainly in the spleen but also in the neck, armpit and groin.

### Monocyte

White blood cells that enter the body's tissues to attack invading organisms and help combat infections.

### Noonan syndrome

Syndrome characterised by unusual facial features, short stature, skeletal anomalies, heart defects, mild intellectual handicap, and bleeding disorders. The syndrome is caused by a mutation in the PTPN 11 gene. Up to 35% of patients with JMML have a mutation in the PTPN 11 gene. In JMML occurring in patients with Noonan syndrome, the cancer is often benign and transitory.

### Philadelphia chromosome (BCR-ABL1)

Chromosome abnormality, also termed BCR-ABL1, which stands for Breakpoint Cluster Region-

Abelson Murine Leukaemia Viral proto-oncogene 1). BCR-ABL1 is a cancer gene formed by the fusion of t(9;22) (q34;q11). It is found in all patients with chronic myeloid leukaemia and some patients with acute lymphoblastic leukaemia.

### Platelets

Platelets are one of the types of blood cells which help to stop bleeding.

### Precursor cell

Precursor cells are a type of partially differentiated cell which has the capacity to differentiate into only one cell type (e.g., precursor B-cell or precursor T-cell).

### Prognosis

Reference to how well a patient is expected to respond to treatment based on their individual characteristics at the time of diagnosis.

### Radiation

Release of energy in the form of particles or waves.

### Refractory (leukaemia)

Refractory leukaemia is a

# Glossary (cont.)

leukaemia that does not result in a remission or that gets worse within six months of the last treatment. However, the leukaemia may be stable.

## Relapse

Relapse occurs when a patient initially responds to leukaemia therapy but, after six months or more, response stops. This is also sometimes called a recurrence.

## Remission

Remission occurs when the following conditions are:

- Blood cell counts returned to normal
- Less than 5% of blasts (early stem cells) are still present in the bone marrow

- There is no leukaemia present elsewhere in the body

## SHP-2

Src homology phosphotyrosine phosphatase 2 is a protein involved in multiple cell signalling processes.

## STAT5

Signal Transducer and Activator of Transcription 5 which are proteins involved in cell signalling.

## Targeted therapy

Drugs that specifically interrupt the ability of the leukaemia to grow in the body. These drugs do not simultaneously harm healthy tissue the way conventional chemotherapy agents do.

## Tell us what you think!

If you would like to give us some feedback about this patient information booklet, please hover over the code to the right using your phone or tablet's camera. Click the link as it appears and this will take you to a short web form to fill in.

Suitable for Android, iPhone 7 and above.



# Useful contacts and further support

There are a number of helpful sources to support you during your diagnosis, treatment and beyond, including:

- Your haematologist and healthcare team
- Your family and friends
- Your psychologist (ask your haematologist or CNS for a referral)
- Reliable online sources, such as Leukaemia Care
- Charitable organisations

There are a number of organisations, including ourselves, who provide expert advice and information.

## Leukaemia Care

We are a charity dedicated to supporting anyone affected by the diagnosis of any blood cancer.

We provide emotional support through a range of support services including a helpline, patient and carer conferences, support group, informative website, one-to-one buddy service and high-quality patient information. We also have a nurse on our help line for any medical queries relating to your diagnosis.

Helpline: **08088 010 444**  
**[www.leukaemiacare.org.uk](http://www.leukaemiacare.org.uk)**  
**[support@leukaemiacare.org.uk](mailto:support@leukaemiacare.org.uk)**

## Bloodwise

Bloodwise is the leading charity into the research of blood cancers. They offer support to patients, their family and friends through patient services.

**020 7504 2200**  
**[www.bloodwise.org.uk](http://www.bloodwise.org.uk)**

## Cancer Research UK

Cancer Research UK is a leading charity dedicated to cancer research.

**0808 800 4040**  
**[www.cancerresearchuk.org](http://www.cancerresearchuk.org)**

## Macmillan

Macmillan provides free practical, medical and financial support for people facing cancer.

**0808 808 0000**  
**[www.macmillan.org.uk](http://www.macmillan.org.uk)**

## Maggie's Centres

Maggie's offers free practical, emotional and social support to people with cancer and their families and friends.

**0300 123 1801**  
**[www.maggiescentres.org](http://www.maggiescentres.org)**

## Citizens Advice Bureau (CAB)

Offers advice on benefits and financial assistance.

**08444 111 444**  
**[www.adviceguide.org.uk](http://www.adviceguide.org.uk)**

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Leukaemia Care is a national charity dedicated to providing information, advice and support to anyone affected by a blood cancer.

Around 34,000 new cases of blood cancer are diagnosed in the UK each year. We are here to support you, whether you're a patient, carer or family member.

## Want to talk?

Helpline: **08088 010 444**

(free from landlines and all major mobile networks)

Office Line: **01905 755977**

**[www.leukaemiacare.org.uk](http://www.leukaemiacare.org.uk)**

**[support@leukaemiacare.org.uk](mailto:support@leukaemiacare.org.uk)**

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**Leukaemia Care**  
YOUR Blood Cancer Charity

