Acute Megakaryoblastic Leukaemia (AMKL)

A Guide for Patients

Leukaemia Care
YOUR Blood Cancer Charity

Introduction

Being diagnosed with Acute megakaryoblastic leukaemia (AMKL) can be a shock, particularly when you have never heard of it. If you have any questions about AMKL, including what causes it, who it affects, how it affects your body, what symptoms to expect and likely treatments - this booklet covers the basics for you.

The booklet was written by our Patient Information Writer, Isabelle Leach, and peer reviewed by Dr. Steve Knapper, academic clinical haematologist at Cardiff University, and David O'Connor a consultant at GOSH. We are also grateful to our reviewer Jason, whose son was diagnosed with AMKL, for their contribution.

If you would like any information on the sources used for this booklet, please email **communications@leukaemiacare.org.uk** for a list of references.

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In this booklet

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 - 5:30pm weekdays and 7:00pm - 10:00pm on Thursdays and Fridays. 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@leukaemiacare.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.leukaemiacare.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.leukaemiacare.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@leukaemiacare.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.leukaemiacare.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@leukaemiacare.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.leukaemiacare.org.uk/communication-preferences/**

What is AMKL?

Acute megakaryoblastic leukaemia (AMKL) is a rare subtype of acute myeloid leukaemia (AML). It is more common in children than in adults. In children with Down syndrome, AMKL is the most common type of AML.

AMKL is characterised by a presence of ≥20% of megakaryoblasts of which 50% or more are of megakaryocyte lineage, meaning that they have developed from a normal precursor cell called a megakaryocyte. The megakaryocyte is a large cell in the bone marrow which produces the platelets in the blood to prevent bleeding.

AMKL is associated with extensive myelofibrosis of the bone marrow. Myelofibrosis is a reactive and reversible process which occurs with many cancerous and non-cancerous diseases of the bone marrow.

In the 2016 World Health Organization (WHO) classification of myeloid neoplasms and acute leukaemia, acute megakaryoblastic leukaemia is included under three subtypes of AML:

AMKL with chromosome t(1;22) (p13;q13) and gene RBM15-MKL1.

Myeloid proliferations related to Down syndrome: Increase in bone marrow cells, usually megakaryoblasts.

- Transient myeloproliferative disorder: Transient abnormal increase of all bone marrow cells
- Myeloid leukaemia associated with Down syndrome

AMKL, not otherwise specified (NOS).

- Defined as an AML with 20% or more blasts, of which 50% or more are of megakaryocyte lineage (descended from the same cell line)

The 2016 WHO classification is based on the findings from immunophenotyping, chromosome analysis, biological features and clinical symptoms. Immunophenotyping is a process that uses antibodies to identify cells based on the types of antigens or markers on the

surface of the cancers cells.

What causes AMKL?

AMKL can occur for the following reasons:

- As a new disease
- Due to a 'secondary effect' of previous chemotherapy treatment
- As a progression from myeloproliferative cancer or myelodysplastic syndrome.

A myeloproliferative cancer is a disease of the bone marrow in which excess cells are produced.

Myelodysplastic syndrome is a disorder in which the blood cells in the bone marrow do not mature to become healthy blood cells.

The exact mechanism(s) for the secondary cases of AMKL are unknown.

The causes of the three distinct subtypes of AMKL are:

- **Translocation of chromosome t(1;22)(p13;q13) and gene RBM15-MKL1:** A chromosome translocation is the transfer of one part of a chromosome to another part of the same or a different chromosome, resulting in rearrangement of the genes.
- **Myeloid proliferations related to Down syndrome:** In children with Down syndrome, the chromosome abnormality trisomy 21 (an extra chromosome 21), which is responsible for Down syndrome, is also thought to be directly involved in the cancerous proliferations of haematopoietic cells, including megakaryoblasts. In addition, nearly all children with AMKL and Down syndrome have a mutation in the GATA-binding factor 1 (GATA1) gene.
- **AMKL, NOS:** This includes other chromosome abnormalities not included in the two subtypes above.

Who is affected by AMKL?

AMKL is a rare subtype of AML, and accounts for 3-5% of all AML cases.

AMKL consists of three distinct

What is AMKL? (cont.)

groups which have differing ages of presentation, causes, responses to treatment and prognoses. These groups are:

- Children with AMKL
- AMKL patients associated with Down syndrome
- Adults with AMKL

AMKL in adults is very rare and represents less than 1% of all individuals diagnosed with AML. In children, AMKL accounts for between 4% and 15% of AML patients.

Children with AMKL can be subdivided as:

- Children with AMKL and without Down syndrome
- Children with AMKL and Down syndrome: These children have a 500 fold increased risk of developing AMKL compared with children without Down Syndrome. However, children with AMKL and Down syndrome do have a better prognosis.

Signs and symptoms of AMKL

Patients with AMKL may be affected in several different ways and may develop one or more of the following disease features:

- Anaemia
- Bleeding or bruising due to thrombocytopenia (low platelet count)
- Increased susceptibility to infections
- Enlarged organs, mainly the liver and spleen
- Bone lesions
- Leukocytosis (increase in the number of white cells in the blood)
- Myelofibrosis (formation of fibrous tissue within the bone marrow that disrupts blood cell production)

Differences in presentation

- Myelofibrosis may be detected before a clear diagnosis of AMKL has been made, or as a late side effect in patients who have already received treatment for leukaemia. Because of the disruption to normal blood cell production by the myelofibrosis, patients may experience tiredness and shortness of breath (due to anaemia), pain below ribs on the left side (due to enlarged spleen), easy bruising/bleeding (due to low platelets), fever and night sweats.
- While some patients with AMKL may present with thrombocytopenia (low levels of platelets) or bone marrow failure associated with pancytopenia (low levels of red cells, white cells, and platelets), thrombocytosis (excessive number of platelets in the blood) may also be present. Patients with thrombocytosis rarely have any signs or symptoms and it is usually picked up in a routine blood test.
- Central nervous system (the brain and spinal cord) involvement is unusual in patients with AMKL, but invasion of the meninges (three membranes that enclose the brain and spinal cord) by

myeloblasts or chloromas (lumps of soft tissue made of myeloblasts), has been described.

- Cases of AMKL caused by the translocation t(1;22) often have an atypical presentation with extramedullary disease (disease located outside the bone marrow), with relatively few blasts in the blood but extensive bone marrow fibrosis.

How is AMKL diagnosed?

The diagnostic criteria for AMKL is a proliferation of ≥20% megakaryoblasts and ≥50% of blasts of megakaryocytic lineage as seen from bone marrow aspirates or peripheral blood.

Peripheral blood

Peripheral blood may contain megakaryoblastic fragments and small blast cells which tend to have variable shapes.

Bone marrow aspirate and biopsy

A bone marrow biopsy is vital for establishing a diagnosis. It involves the collection of a sample of bone marrow from the hip bone, generally under local anaesthesia (but usually done with a general anaesthetic in children). This generally consists of two types of samples known as an aspirate and a trephine that are taken during the same procedure.
A bone marrow biopsy needle with a cylindrical blade, called a trephine, is used to remove a 1 or 2cm core of bone marrow in one piece.

A bone marrow aspirate, which consists of taking a sample of the liquid part of the soft tissue inside your bones using a syringe is less invasive, but in patients with AMKL where there is extensive myelofibrosis, the bone marrow aspirate is often very slight, and a diagnosis cannot always be achieved. In children with AMKL, the bone marrow aspiration may be easier to perform, particularly in children with Down syndrome.

The aspirate or biopsy samples are examined under the microscope to determine the number and type of cells present and the level of haematopoiesis (the process by which blood cells are formed). Bone marrow biopsies can show numerous blasts, clusters of micro-megakaryoblasts or more mature megakaryoblasts. There is a corresponding decrease in usual bone marrow maturation. Cytoplasmic blebs may also be identified to help with the diagnosis.

A definitive diagnosis of AMKL can be reached when the criteria set out in the WHO 2016 classifications for myeloid neoplasms and acute leukaemia

are met for any of the three AMKL subtypes previously mentioned:

- AMKL with chromosome t(1;22) (p13;q13) and gene RBM15-MKL1
- Myeloid proliferations related to Down syndrome
- AMKL, NOS

The proliferation of ≥20% megakaryoblasts can be documented by the use of flow cytometry. This technique documents the number of megakaryoblasts by identifying their specific antigens, separating the different types from blood or bone marrow, and then counting them.

When flow cytometry cannot be performed due to absence of a bone marrow aspirate sample as in the case of extensive myelofibrosis, immunophenotyping can sometimes be performed on atypical (unrepresentative) cells isolated from the bone marrow or blood to make a diagnosis.

Immunophenotyping is a process that uses antibodies to identify the types of antigens or markers on the surface of the cells. It is used to diagnose specific types of leukaemia. In cases of AMKL, the cells normally show positivity for CD33, CD13, CD41, CD42, CD61 and factor VIII, and negativity for myeloperoxidase, an enzyme produced by certain types of white blood cells.

Further evidence to confirm the diagnosis comes from chromosome analysis which is based on the patient group affected by AMKL.

Children with AMKL

Population characteristics

- Occurs in children younger than three years of age without Down syndrome, most commonly in females.

Causes

- **Translocation of chromosome t(1;22)(p13;q13) and gene RBM15-MKL1:** A chromosome translocation is the transfer of one part of a chromosome to another part of the same or a different chromosome, resulting in rearrangement of the genes.
- **AMKL, NOS:** This subtype includes chromosome abnormalities other than those that feature in the subtypes of 'translocation of chromosome t(1;22)(p13;q13) and gene RBM15-MKL1' and 'myeloid leukaemia associated with Down syndrome.'

Treatment

Treatment for patients with AMKL will depend to a large extent on the subtype they have been diagnosed with, as well as their clinical, immunophenotyping and chromosomal abnormality features.

Despite recent improvements in the understanding of the causes of AMKL, the optimal treatment is still debated. Currently, there is no 'targeted therapy' available for AMKL. Targeted therapy refers to drugs that specifically interrupt the leukaemia cells from growing in the body. These drugs do not simultaneously harm healthy cells the way conventional chemotherapy drugs do.

Some haematologists treat children with AMKL but without Down syndrome as very high-risk, and recommend an allogeneic stem cell transplantation (ASCT) as soon as complete remission has been achieved. Other haematologists treat these children with intensive chemotherapy only, and excellent survival rates, which haven't been bettered with ASCTs, have been achieved with this treatment.

The standard of care treatment protocols for AMKL, which come from a number of international trials, differ both in the drug schedules and the intensity of treatment. However, they nearly always include cytarabine and

an anthracycline. Anthracyclines were originally antibiotics first isolated from the fungus Streptomyces peucetius in the 1950s, and subsequently were found to be effective anti-cancer drugs. The protocols are normally tailored to the patients' clinical and genetic characteristics.

In a review study in which children with AMKL received intensive chemotherapy alone, the 10 year overall survival was 76% for patients without Down syndrome and 79% for patients with Down syndrome. Moreover, the study showed that for patients without Down syndrome who did not achieve remission, ASCT did not offer any advantage over chemotherapy alone.

Prognosis

The prognosis for children with AMKL but not Down syndrome is controversial. While some haematologists consider this disease to be high-risk and requiring of immediate treatment, others treat it as standard risk, unless there are chromosomal abnormalities present and/or a poor response to induction therapy.

The prognosis of non-Down Syndrome children with AMKL is worse than that for other forms of AMKL. In the AML-BFM 98 study of 118 patients with AMKL, the three-year survival was 91% in patients with Down syndrome and only 64% in patients without Down syndrome.

In a large review study of 153 non-Down syndrome patients with AMKL treated with several intensive chemotherapy protocols that included cytarabine and an anthracycline, overall four-year survival rate was estimated at 56%.

In a study analysis of the chromosomes of children with AMKL but not Down syndrome, the gene inv(16) CBFA2T3-GLIS2 was present in nearly 30% of these patients and predicted an exceptionally poor outcome, with a five-year survival rate of 34.3% compared with the five year survival rate of 88.9% for the AMKL patients without this gene. This gene would appear to be a useful prognostic factor.

Children with AMKL (cont.)

Conversely, the results of two studies of AMKL patients without Down syndrome suggest that the t(1;22) translocation may indicate patients that could have a favourable prognosis compared to other subtypes of AMKL, provided that intensive chemotherapy and adequate supportive care are provided.

AMKL patients with Down syndrome

Patient characteristics

Transient myeloproliferative disorder:

- This is a temporary abnormal increase of all bone marrow cells which occurs at birth or within a few days of birth, and usually resolves within one to two months. It is found in 10% of newborn infants with Down syndrome.
- Up to 20% of children with transient myeloproliferative disorder go on to develop AMKL within the first four years of life.

Myeloid leukaemia associated with Down syndrome:

- Occurs usually in the first four years of life and requires treatment.
- Average age at diagnosis is 1.8 years.
- White cell counts are often lower in children with Down syndrome associated myeloid leukaemia than in children with AMKL who do not have Down syndrome.

Causes

Transient myeloproliferative disorder:

- The causes of transient myeloproliferative disorder and the reasons for its progression in some children are not fully understood; however, it is thought to be involved with Down syndrome.

Myeloid leukaemia associated with Down syndrome:

- The development of myeloid leukaemia is more common in children with Down syndrome, who have the chromosome abnormality trisomy 21. This chromosome abnormality is thought to be directly related to the cancerous transformation of haematopoietic cells (stem cells that give rise to all types of blood cells), including megakaryocytes.
- Nearly all of the children with myeloid leukaemia associated with Down syndrome have a GATA1 mutation, as well as the trisomy 21 chromosome abnormality.

Treatment

Transient myeloproliferative disorder:

- Treatment with low dose cytarabine (0.5–1.5 mg/kg) for three to 12 days achieved a five-year overall survival rate of 85% in a study of 146 Down syndrome children with transient leukaemia. However, whether treatment of transient myeloproliferative disorder will alter the risk of developing subsequent AMKL is still unclear.

Myeloid leukaemia associated with Down syndrome:

- Children with myeloid leukaemia associated with Down syndrome have better results with intensive chemotherapy compared with patients without Down syndrome, but they are more susceptible to the toxic side effects of the chemotherapy.
- Very good results have been achieved for myeloid leukaemia associated with Down syndrome using dose-reduced treatment protocols.

Children with Down syndrome in the ML-DS 2006 study received a dose-reduced version of the treatment protocol given in the AML-BFM 98 trial which was achieved by reducing the total dose of etoposide, reducing the CNS prophylaxis (cytarabine delivered direct into the spinal fluid by lumbar puncture) and excluding maintenance therapy. The children in the ML-DS 2006 study showed excellent results with a five-year overall survival of 89%, which was similar to the three year survival of 91% for the children with Down syndrome in the AML-BFM 98 trial. In addition, the children in the ML-DS 2006 study showed milder toxicity. These results illustrate that reducing therapy in patients with AMKL and Down syndrome, who are sensitive to toxic side effects of the chemotherapy, does not decrease overall survival.

Prognosis

Overall, patients with AMKL associated with Down syndrome have an excellent prognosis with a greater than 90% overall survival rate.

AMKL patients with Down syndrome (cont.)

Transient myeloproliferative disorder:

- A large international study analysed the characteristics of transient myeloproliferative disorder in 264 children with AMKL and Down syndrome. Overall survival rate was approximately 80%, and the early death that occurred in up to 20% of infants occurred in those with a higher white blood cell count at diagnosis, increased bilirubin (a breakdown product of red blood cells) and liver enzymes, and a failure of the blood cell counts to return to normal with treatment.
- Subsequent development of myeloid leukaemia was seen in approximately 19% of infants after a mean period of 20 months following the occurrence of the transient myeloproliferative disorder.

Myeloid leukaemia associated with Down syndrome:

- Overall survival rates in recent clinical studies of patients with myeloid leukaemia associated with Down syndrome were 80-90%. However, if these patients relapse following chemotherapy, overall survival rate decreased dramatically, and patients had a far poorer outlook.
- In addition to having the chromosome abnormality trisomy 21, the development of myeloid leukaemia was noted to be significantly greater in patients with other chromosome abnormalities including trisomy 11, del 16q, der(14;21), t(5;13), and tetrasomy 21.
- A poor prognosis is associated with an older age at diagnosis.

Adults with AMKL

Patient characteristics

- AMKL in adults occurs in only 1% of all AML cases.
- Median age of adults with AMKL was 56 years (range: 21-78 years) in a study of 37 patients with AMKL.

Causes

- The genetic basis for adults with AMKL is poorly defined given the rarity of the disease.
- AMKL in adults occurs as part of the AMKL, NOS subtype, which includes chromosome abnormalities other than those featured in the subtypes of 'translocation of chromosome t(1;22)(p13;q13) and gene RBM15-MKL1' and 'myeloid leukaemia associated with Down syndrome'.
- AMKL may also progress from myeloproliferative cancer or myelodysplastic syndrome.

Treatment

Intensive chemotherapy is a necessity for adults with AMKL but not Down syndrome because they have a very poor prognosis. In a study of 37 patients with AMKL, complete remission rates were 43% and overall survival was 23 weeks.

Prognosis

From the limited experience of AMKL in adults, a poor prognosis, despite treatment with intensive chemotherapy, can be expected but there is still room for hope.

Although the overall survival rate is around four to ten months, complete remission is achieved in around half of adult AMKL patients.

Living with AMKL

Because AMKL occurs mainly in children and AMKL in adults only represents 1% of all cases, the following sections are written for the parents of a child diagnosed with AMKL. However, all the advice is equally applicable to adults with AMKL or the relatives of an adult with AMKL.

Emotional impact of AMKL

Being told your child has cancer can be very upsetting. Seeing your child with some of the symptoms of AMKL 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 AMKL, 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 for them to try and stay as active as possible to make them 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 AMKL 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.
- Taking part in regular, gentle play will help 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 their doctor or nurse.

Practical support

Work and finances

If your child is diagnosed with AMKL, 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 AMKL 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 their nurse in the hospital how to apply.

Talking about AMKL

Talking to the haematologist

AMKL is a rare condition. It is important for you to develop a good working relationship with your child's 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 AMKL.

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 AMKL.

Talking to other people

Telling people that your child has a rare condition like AMKL 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 or booklets about AMKL like this one 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

Talking about AMKL (cont.)

produces

- Explain your child's symptoms (maybe they are tired, or have a lot of pain)
- Explain what you 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 your 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.

Glossary

Allogeneic stem cell transplant (ASCT)

A stem cell transplant of cells from a matching donor.

Anaemia

A condition where the number of red blood cells, which contain haemoglobin and transport oxygen to body cells, are reduced. This may be due to a lack of iron anaemia, leukaemia or sickle cell disease.

Antibody

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

Antigen

A toxin or other foreign substance which induces an immune response in the body, especially the production of antibodies.

Bilirubin

The breakdown product of red blood cells.

Bleb

A bulge or protrusion of the plasma membrane of a cell.

Central nervous system

Consists of the brain and spinal cord.

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.

Chloroma

Soft tissue mass consisting of myeloblasts which typically occurs in patients with acute myeloid leukaemia. The chloroma is located outside of the bone marrow and can be found anywhere in the body.

Chromosome Inversion

Chromosome inversion occurs when a segment is clipped out, turns upside down, and reinserts itself back into the chromosome. This may be inherited or a random mutation.

Cytokine

Any of a number of substances, such as interferon, interleukin, and growth factors, which are secreted by certain cells of the immune system and have an effect on other cells.

DNA (deoxyribonucleic acid)

A thread-like chain of amino acids found in the nucleus of each cell in the body which carries genetic instructions used in the growth, development and functioning of the individual's cells.

Epidemiology

The study of how often diseases occur in different groups of people and why.

Extramedullary disease

A disease located outside the bone marrow.

GATA1 (GATA binding protein 1) gene

This gene generates directions on how to make the proteins that bind to specific regions of DNA and help regulate many other genes. Particularly, GATA1 gene is essential for the maturation and controlled death of megakaryocyte cells.

Genes

Genes are made up of DNA which stores the genetic information required to make human proteins.

Haematopoiesis

The process by which blood cells are formed.

Immunophenotyping

The process that uses antibodies to identify cells based on the types of antigens or markers on the surface of the cells. This process is used to diagnose specific types of leukaemia and lymphoma by comparing the cancer cells to normal cells of the immune system.

JAK gene

Janus Kinase gene which manages signals from cytokines.

Lineage

Descended from a specific cell line.

Megakaryocyte

Large cell in the bone marrow which produces the platelets in the blood to prevent bleeding.

Meninges

Three membranes, known as the dura mater, arachnoid mater, and pia mater, that enclose the brain and spinal cord.

Myelofibrosis

Reactive and reversible process which occurs with many cancerous and non-cancerous diseases of the bone marrow.

Glossary (cont.)

Myeloid

Relates to bone marrow.

Myeloproliferative cancer

A disease of the bone marrow in which excess cells are produced.

Prognosis

Indication of how well a patient is expected to respond to treatment based on their individual characteristics at the time of diagnosis or other timepoint in the disease.

Proliferation

Rapid increase, for example in the number of cells.

STAT genes

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

Targeted therapy

Drugs that specifically interrupt the leukaemia cells from growing in the body. These drugs do not simultaneously harm healthy cells the way conventional chemotherapy drugs do.

Thrombocytopenia

Low levels of platelets, which are small blood cells that help the body form clots to stop bleeding.

Thrombocytosis

Excessive number of platelets which are small blood cells that help the body form clots to stop bleeding.

Translocation

In genetics, translocation is the transfer of one part of a chromosome to another part of the same or a different chromosome, resulting in rearrangement of the genes.

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
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

Cancer Research UK

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

0808 800 4040
www.cancerresearchuk.org

Macmillan

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

0808 808 0000
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

Citizens Advice Bureau (CAB)

Offers advice on benefits and financial assistance.

08444 111 444
www.adviceguide.org.uk

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

support@leukaemiacare.org.uk

Leukaemia Care,
One Birch Court,
Blackpole East,
Worcester,
WR3 8SG

Registered charity
259483 and SC039207

Leukaemia Care
YOUR Blood Cancer Charity

Health & care information you can trust
The Information Standard Certified Member