RUNX-101: An Adolescent's Guide to Understanding and Communicating about RUNX1-Familial Platelet Disorder (FPD)

National Institutes of Health
Turning Discovery Into Health

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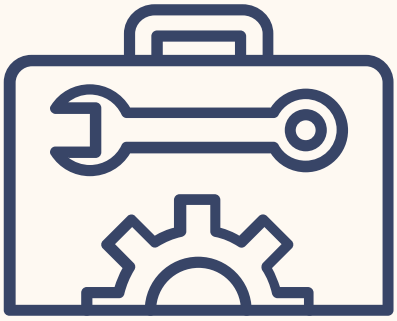
A Toolkit of Information

Living with a medical condition, especially a rare one, can be challenging. This guide aims to help you better understand *RUNX1* and the role it plays in your health. You can think of this as your personal toolkit: pull out specific sections of information that may be helpful to you at any particular moment or read the content in its entirety if that works best for you.

This guide should not substitute discussion with your healthcare providers. If you have questions, it can be helpful to write them down to ask your medical team. They are always available to support you.



Note**:** Sometimes people italicize *RUNX1,* and sometimes it's just written RUNX1. This is because in genetics we italicize the names of genes, but if we are referring to the protein that gene ultimately produces or the resulting medical condition then it is written in standard text. Any word bolded is defined in the glossary.

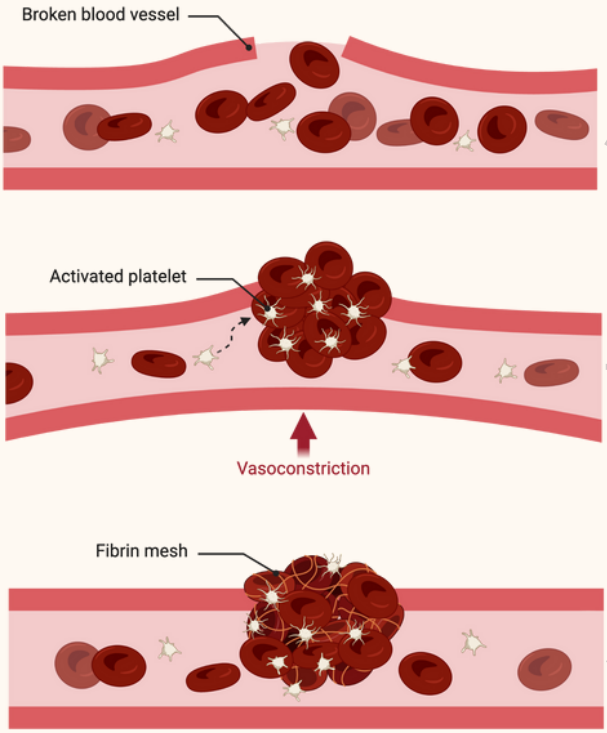
The Fundamentals of RUNX1-Familial Platelet Disorder (FPD)

Q1. What is blood made up of?

Blood is made up of lots of different types of cells, including red blood cells, white blood cells, and our favorite: platelets. These cells are generated from a spongy area inside our bones called bone **marrow.**

- ***Red blood cells*** *transport oxygen around the body*
- ***White blood cells*** *fight against infections and build immunity*
- ***Plasma*** *is the liquid that transports blood cells and other molecules throughout the body*

Platelets function like internal "Band-Aids" that thicken blood when we get wounded to protect against excessive bleeding.



1. *Blood vessel is damaged*
2. *Platelets rush to injury and form sticky web that pools red blood cells together (fibrin mesh)*
3. *This stops more bleeding until vessel heals*

The Fundamentals of RUNX1-Familial Platelet Disorder (FPD)

Q2. I've heard that RUNX1-FPD is "genetic." What are genes?

We have ~20,000 genes which make thousands of different proteins, each with their own function!

Genes tell our body what it needs to know to function and grow. Genes code for specific products called **proteins** that have various functions, such as making our eyes a certain color or helping us move.

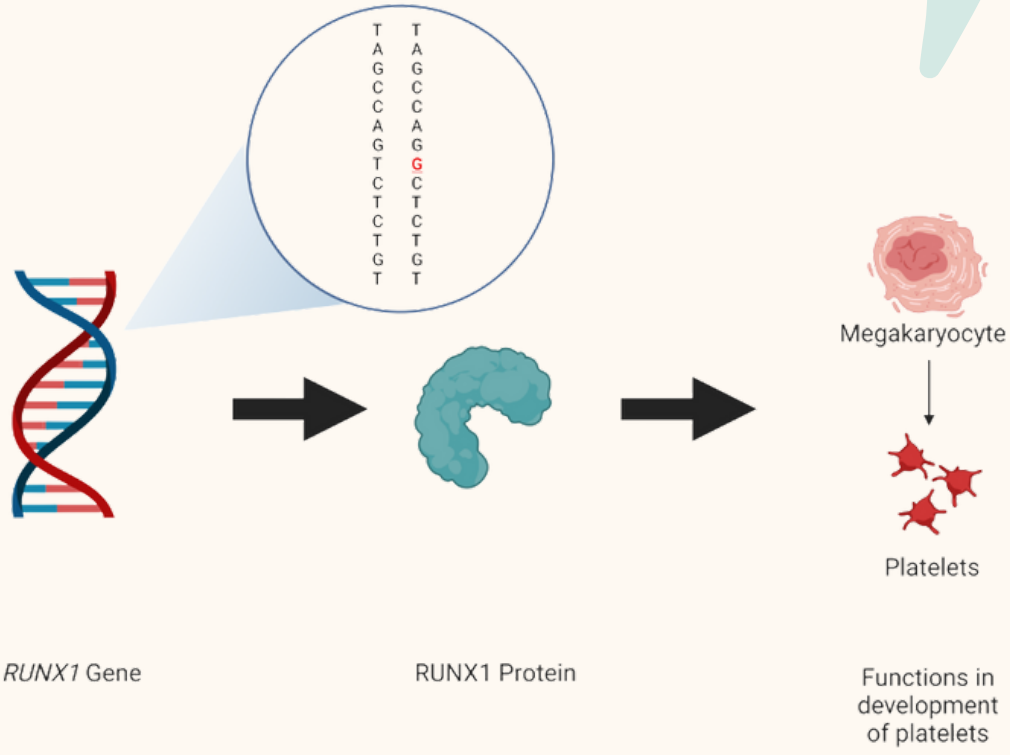
Genes are also made up of DNA and are written in a code (using the letters A,T,C & G) just like words in a book. Our **genome** (all the genes in our body) is almost 3.4 billion letters long! We have two copies of almost every gene in our body, and we get one copy from each of our parents.

As you can imagine, a 3.4 billion-letter book is bound to have a few typos. We refer to typos in DNA as genetic variants (or mutations). Genetic variants happen all the time. They aren't always bad; we all have thousands of genetic differences that make us unique. However, when a genetic variant impacts a gene with an important role in health, it can lead to a **genetic condition.**

The Fundamentals of RUNX1-Familial Platelet Disorder (FPD)

Q3. What is the RUNX1 gene?

The RUNX1 gene codes for the RUNX1 protein. RUNX1 is a **transcription factor,** which means it helps to turn other genes on and off when the body needs them. The RUNX1 protein turns on genes that help control the development of blood cells (hematopoiesis). In particular, it plays an important role in development of early blood cells that can develop into all types of mature blood cells such as white blood cells, red blood cells, and platelets.



The Fundamentals of RUNX1-Familial Platelet Disorder (FPD)

Q4. Why do I have RUNX1-FPD?

We all have two copies of the RUNX1 gene (one from each of our parents). However, if there is a **pathogenic** (disease-causing) genetic variant in one of these copies, the body may not read the genetic recipe correctly. This leads to the RUNX1 protein not being built correctly, so the platelets do not develop correctly either.

Since genes like RUNX1 are shared between family members, genetic conditions like RUNX1-FPD can be passed on to family members without our control. The term familial refers to medical conditions that are shared with other family members through genetics.

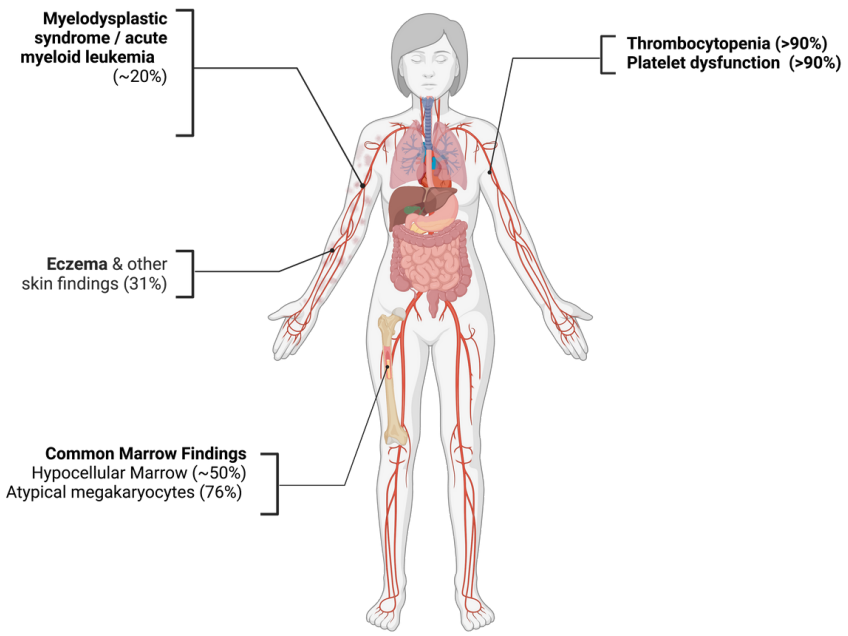
The Fundamentals of RUNX1-Familial Platelet Disorder (FPD)

Q5. What are the symptoms and characteristics of RUNXT-FPD?

RUNX1-FPD primarily affects the platelets. People with RUNX1-FPD could have thrombocytopenia (too few platelets) and the platelets they do have may not work correctly. This causes primary symptoms like easy bleeding and bruising.

Genetic conditions like RUNX1-FPD are NOT contagious, so you don't have to worry about "spreading" the symptoms to friends.

Not everyone will have or express the same symptoms– even in families with the exact same RUNX1 variant. This is called variable expressivity. Some people may have more severe bleeding and bruising while others will have less, and only some people will go on to develop blood cancer or leukemia.

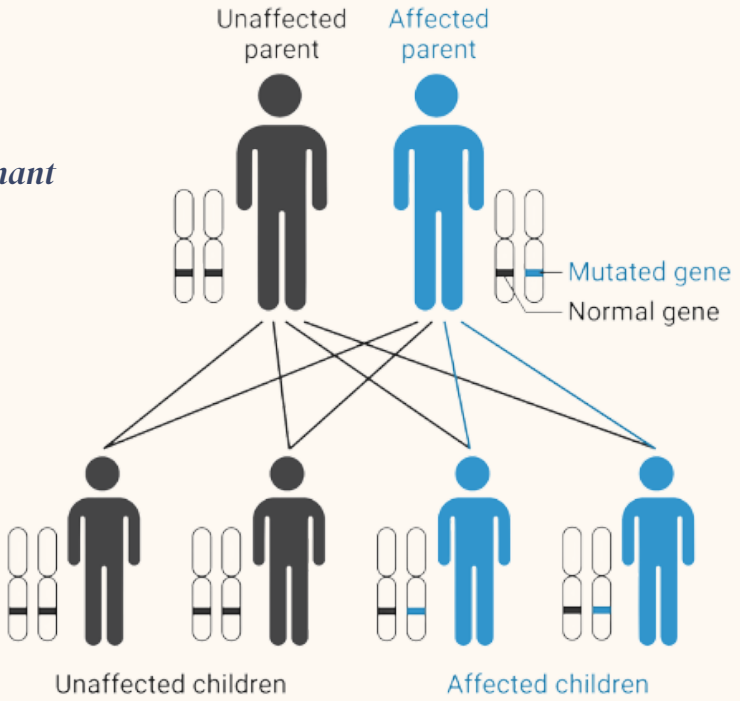


The Fundamentals of RUNX1-Familial Platelet Disorder (FPD)

Q6. How does RUNX1-FPD get passed on through families?

RUNX1-FPD is inherited in an **autosomal dominant** manner. People with RUNX1 have a 50-50 (or 1 in 2) chance of passing on the copy of their RUNX1 gene with the **pathogenic** (disease-causing) genetic variant on to a child. Only children who inherit the variant will be able to pass it onto the next generation. If you have questions/concerns about having a child in the future, you can reach out to a genetic counselor or another health provider such as a gynecologist for additional advice.

Autosomal dominant inheritance



The Fundamentals of RUNX1-Familial Platelet Disorder (FPD)

Q7. Neither of my parents have RUNX1. How is that possible?

We inherit most of our genetic variants from a parent, but sometimes variants can start in a person for the very first time. This happens in the early stages of embryonic development when the DNA is being copied over from the parent's DNA for the very first time. We call a variant that is not inherited from either parent **de novo** (Latin for 'from the beginning').

The average person will have ~50 de novo variants that neither of their parents had. Usually these happen in a way that does not lead to disease, but they can lead to disease if important genes like RUNX1 are involved.

Once someone has a genetic variant, they can pass it on to their children. This means that a person with a de novo RUNX1 variant will still have a 50-50 chance of passing it on to a child (see Q6).

The Fundamentals of RUNX1-Familial Platelet Disorder (FPD)

Q8. What about cancer risk?

While most people with RUNX1-FPD will experience low platelet count and easy bleeding and/or bruising, some people will go on to develop a blood cancer such as **Acute Myeloid Leukemia (AML)** or **myelodysplasia (MDS).** It is estimated that somewhere between 30% to 50% of people with RUNX1-FPD will develop a blood cancer at some point in their life. While 30% may feel like a really large number, another way to look at it is that there's a 70% chance you won't develop blood cancer.

Tell your parents or doctor if you notice any changes in your health – such as feeling abnormally tired, losing weight quickly, or getting frequent fevers/ infections.

While knowing that you are at a higher risk of developing cancer can be scary, having this information gives your medical team the ability to better monitor you so that if a blood cancer starts to develop, it can be caught and treated early. Your doctor may want to do some tests to look at your blood cells and bone marrow to check for early signs of disease. It is important for you to be proactive about your doctor's visits and blood tests to make sure you stay as healthy as possible.

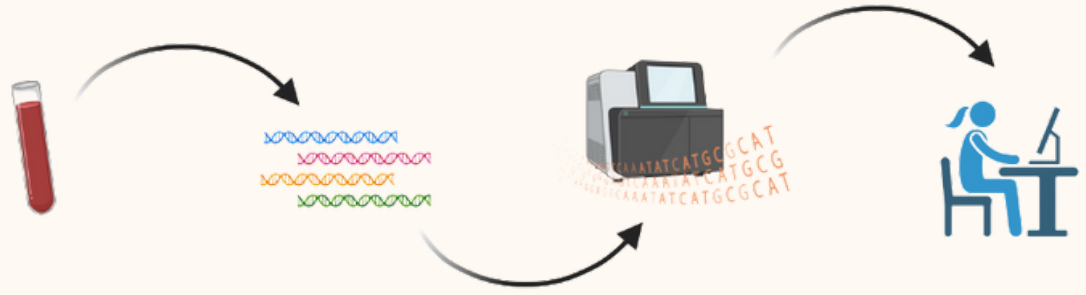
It may be helpful to also talk to a medical professional such as a genetic counselor or hematologist (a doctor who focuses on blood disorders) about cancer risk.

RUNX1-FPD Treatment and Management

Q9. How is RUNX1-FPD diagnosed?

RUNX1-FPD is diagnosed through **genetic testing** (sometimes called sequencing), which is like running spell-check on your genes. There are many different types of genetic testing. Some tests will just look at the RUNX1 gene, or even just a single RUNX1 pathogenic variant that was detected in another family member. Other genetic tests can look at all 20,000 genes at once! Sometimes your parents' DNA will be tested too.

DNA is extracted from blood (or saliva or skin) and run through a machine which prints out a genetic code using the letters A,T,C and G. A scientist in the lab then analyzes that data to look for any variants



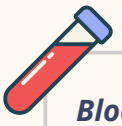
Check out the following resources for more information about the different types of sequencing in genetic testing:

- **https://www.cdc.gov/genomics/gtesting/genetic_testing.htm**
- **https://www.thermofisher.com/us/en/home/life-science/sequencing/sequencing-learning-center/sequencing-basics/dna-sequencing-technologies.html**
- **https://www.genome.gov/genetics-glossary/DNA-Sequencing**

RUNX1-FPD Treatment and Management

Q10. What do I need to do to manage my RUNX1-FPD and stay healthy?

Once we know that a person has RUNX1-FPD, it's important to make sure that they are taken care of as best possible. Everyone's monitoring plan will be a bit different depending on personal, family, and medical history. Typically monitoring will include some combination of visits to a primary care doctor, a hematologist, as well as blood draws and potentially a bone marrow biopsy.



Blood draw:

- *Allows doctors to see if there are any changes in your blood Recommended*
- *every 3 to 6 months*
- *If needle pain is a concern, you can ask your doctor for numbing cream*

Bone marrow biopsy (aspirate):

- *Uses needle to collect bone marrow from hip, checks overall health of bone marrow*
- *May feel sting from needle, temporary tingling in leg during the procedure, and mild soreness/bruising after the procedure*
- *Recommended every one to two years*

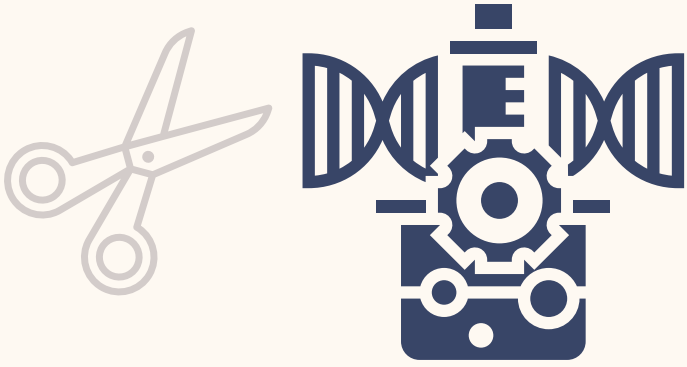
While we talk a lot about taking care of all the medical elements of RUNX1-FPD, it is also important to take care of yourself emotionally, such as making time to do fun activities and practicing self-care. It's okay to ask for help if you are feeling stressed, anxious, or sad. You can also check out our wellness guide for breathing activities to ease any anxiety before a blood draw or bone marrow biopsy.

RUNX1-FPD Treatment and Management

Q11. Is there a cure?

Right now, there is no cure for RUNX1-FPD. However, there are ways to control the symptoms associated with it. For example, medications can be used to help to stabilize clots and control bleeding. While you wouldn't want to take these medications every day, you can take them before a surgery or event involving potential bleeding/ bruising.

There is a lot of exciting research going on in the RUNX1 space. Scientists are looking at new therapies and even using technologies like gene editing or **CRISPR** to try to treat RUNX1-FPD. We hope that soon there will be better treatments, if not a cure, for this condition. Research depends on people with RUNX1-FPD volunteering to participate in studies. For more information on participating in research, see Q19.



RUNX1-FPD Treatment and Management

Q12. If I develop blood cancer, what next?

Your medical team will be your go-to source of information about your treatments and your medical plan. Treatment for blood cancer may include a combination of chemotherapy and possibly a **hematopoietic stem cell transplant (HCT) or bone marrow transplant (BMT).**

With an HCT or BMT, some early-stage blood cells are taken from a donor (sometimes a family member) to replace the cancerous blood cells in the person being treated and restore the two normal copies of the RUNX1 gene in their blood cells.

Transplants can come with unpleasant side-effects, but new types of transplants are being developed to minimize them. The following resources may be helpful for you to learn more:

1. https://www.canteen.org.au/resource/a-young-persons-guide-to-dealing-with-cancer/
2. https://www.amazon.com/Neutrophils-Guide-Stem-Cell-Transplants/dp/0985373644

Getting diagnosed with blood cancer can be scary and frustrating. It may be helpful to connect with other patients with blood cancer or to talk to a healthcare professional (such as a counselor) to help you work through some of the very natural emotions that come with a diagnosis.

Consider asking your medical team some of the following questions:

- *"What type of cancer do I have? What is my exact diagnosis?"*
- *"What is the treatment plan? What are the goals of my treatment? What are the side effects I can expect from them?"*
- *"How do these treatments work? Will they make me feel sick?"*
- *"What are some things I can do to help make these treatments easier"*
- *"Are there other people I can talk to who are going through or have gone through the same treatments as me?"*

Talking About RUNX1

Q13. Why and what should I share?

If you are doing something where there's a higher chance of bleeding, it is important for some people to know that you have RUNX1-FPD so that they can support you if needed. You should also let all of your doctors and dentists know about your RUNX1-FPD to make sure that you get the best medical care. The following responses look into communicating with teachers/coaches, parents, and friends.

Q14. How do I share this with my teacher/coach?

- Try to reach out at the beginning of the school year (if your parents haven't already).
- If privacy is important to you, set aside a time when other students are not there.
- Share the basics about your condition, your symptoms, and any limitations (medical, physical, psychosocial) that you want them to know.
- If you play sports, work with your coach and doctor on a plan to help you continue playing safely.

Talking About RUNX1

Q15. How do I talk about RUNX1-FPD with my parents?

If you feel like you can't openly communicate with your parents the way you want to about your concerns, there are other professionals that can answer questions or concerns, such as genetic counselors and therapists. See page 22 for more information about your RUNX1-FPD team.

While it may not always feel like something you want to do, keeping your parents updated on how you're feeling (physically and emotionally) can create a stronger support system. Since a parent could also share the mutation (see Q7 for exceptions), they may have experience with what you are going through.

Q16. How do I talk about RUNX1-FPD with my friends?

While keeping your diagnosis private is completely up to you, talking to your trusted friend(s) about your condition can be a valuable form of support. Here are a few examples of ways to open the conversation/address their questions:

- "Have you ever noticed that I often have bruises on my body? Well, I learned it is because I have a genetic condition called RUNX1-FPD."
- "Don't worry, RUNX1-FPD isn't contagious, and you won't get it from hanging out with me."
- "That genetics lecture in bio class really reminded me of my own experience with RUNX1-FPD."

Talking About RUNX1

Q17. How does this apply to my siblings?

While you and your siblings inherit a lot of the same genetic material from your parents, there are two copies of every gene, and your siblings can inherit different copies. This means that your sibling(s) may not inherit the same RUNX1 variant and may not develop the condition. While it may seem unfair that you must go to the doctor more than your siblings, this is for your own wellbeing.

Your siblings may have their own reactions to your diagnosis and struggle to process their own emotions. They could have questions for you and your parents. Try to be open with your siblings when they do have questions, but also know it's okay to ask them for space when you don't feel comfortable talking about things.

Talking About RUNX1

Q18. How can I connect with others who have RUNX1-FPD?

A lot of people find it helpful to connect with other people their age with RUNX1-FPD. Your doctors or NIH Natural History Study providers may be able to connect you with another patient in your age range.

Some people with RUNX1-FPD might find it helpful to connect with others on social media who are going through the same things as them. If you do choose to use social media:

- It is up to you to decide what and how much you feel comfortable sharing
- Connections made or information from social media should not replace conversations with your medical team
- Monitor your own emotions while using social media groups that discuss difficult topics
- Take breaks and try to differentiate the context of other people's posts from that of your own life
- Always check the privacy settings of a group before posting personal medical information - even private messages can be saved as screenshots

RUNX1-FPD and Research

Q19. Should I participate in research?

Scientists at the NIH and beyond are very interested in learning more about RUNX1-FPD and different types of blood cancers. There are research studies that patients with RUNX1-FPD can join, such as the NIH RUNX1 Natural History Study– you may already be enrolled! The goal of these studies is to understand the biology of the disease and hopefully to develop better ways of caring for families like yours.

Some people find it really rewarding to participate in research and learn about their condition. However, participating in research can be a big commitment. For any study you consider, you should speak with an investigator about the benefits, risks and what participation could look like for you.

Takeaways:

- While your individual feelings and concerns may be unique, you are not alone
  - Connecting with others with RUNX1-FPD can help reduce your feelings of isolation.
- Having RUNX1-FPD isn't anyone's fault
  - It is a genetic and (usually) hereditary condition.
- Everyone copes differently
  - Your feelings may change over time; instead of keeping your feelings inside, it may be helpful to share them with someone you trust and to acknowledge those feelings as a part of the process.
- Ask questions
  - Seeking help and getting the right information is important. You can write your questions and answers down on the next page about your RUNX1-FPD team.

Additional Resources:

READINGS:

- *Child Wellness Toolkit: A Caregiver's Guide*
  - Helpful wellness/coping resources for people of all ages, such as the Sources of Strength section with links to coping activities, feel-good music playlists and more.
- *Paving the Road: A RUNX1 Communication Guide for Parents*
  - Communication guide for parents initiating a conversation about RUNX1-FPD with their child, with resources in the Middle/High school section.

ORGANIZATIONS:

- National Society of Genetic Counselors (NSGC) Find A Genetic Counselor tool
  - If you haven't already met with a genetic counselor, you can ask your parents about trying to connect with one either in person or online. This resource is a good place to start.
- The Leukemia and Lymphoma Society Young Adult Resources
- National Organization for Rare Diseases
- The National Human Genome Research Institute Glossary of Genetics Terms

Your RUNX1-FPD Healthcare Team

This table helps organize all the information about who is looking after you. You may not interact with all of these professionals or have them all on your team, and one professional may have multiple roles. You can fill in the names and contact information of the healthcare professionals on your team.

Professional	What They Do	Name and Contact
Genetic Counselor	Uses family history to assess individual/family risk of a heritable condition (like RUNX1)	
Hematologist	Specializes in blood diseases	
Nurse	Performs physical exams, carries out plan of care, coordinates with health care team, and provides education and support	
Nurse Practitioner or Physician Assistant	Advanced practice provider that assesses patient needs, interprets diagnostic and laboratory tests, diagnose disease, and prescribes treatments	
Patient Care Coordinator (Navigator)	Organizes patient care activities, connects with other members of the care team, and may coordinate treatments, travel and lodging	

Any other questions you want to ask your healthcare provider/genetic counselor? (eg. school, teachers, communicating with family, any sports precautions, social media groups, concerns about future family etc....) Write them down below:

_____

_____

_____

_____

_____

Other Specialists You May Meet

Allergist	Specializes in allergies	
Dermatologist	Specializes in skin disorders	
Gastroenterologist	Specializes in stomach and intestinal disorders	
Gynecologist	Specializes in female reproductive health	
Neurologist	Specializes in brain and nervous system disorders	
Oncologist	Specializes in tumors and cancer	
Pain Management Specialist	Specializes in acute and chronic pain	
Psychologist	Therapist who help with emotional and intellectual well-being	
Psychiatrist	Medical doctor who, in addition to the role of a psychologist, can prescribe medication	
Pulmonologist	Specializes in respiratory system (breathing and oxygen flow)	
Rheumatologist	Specializes in musculoskeletal and autoimmune conditions like arthritis	
Social Worker	Supports children and families, helps them cope and adapt	

Glossary

A

Acute Myeloid Leukemia and Myelodysplasia — Cancer of blood and bone marrow, interfering with normal production of white blood cells, red blood cells, and platelets

Aminocaproic Acid (Amicar)or other platelet medications— Prescription medicine to help control bleeding when the blood doesn't clot the way it should
This is a way of giving your body some extra band aids to help you heal.

Autosomal Dominant — Pattern of inheritance; gene is located on a non-sex chromosome, and only a single of the variant from a parent is needed to cause the genetic disease
There are certain patterns to how genes (and genetic traits) are shared within a family. "Autosomal" means that the gene in question is located on one of the numbered, or non-sex, chromosomes. "Dominant" means that only one of a person's two copies of a gene needs to be impacted to cause the condition. Each child of a person with an autosomal dominant condition has a 50%, or 1 in 2, chance of also having that condition. RUNX1-FPD is an autosomal dominant condition.

B

Benign— Not disease-causing

Blood Draws— Procedure that collects blood from a vein for testing to see if there are any changes in blood that could be concerning
A procedure to collect blood so that doctors can check how healthy you are. A needle is inserted into the vein of the arm or hand to collect just a few teaspoons of blood for analysis in a lab. Blood draws aren't always fun, but they are important for keeping you healthy. You will feel a little pinch when the needle is first inserted into your vein. Blood draws usually won't last for more than a minute or so. You can always ask your doctor for numbing cream or other tools to help make blood draws not hurt as much.

Bone Marrow— Soft spongy material found inside bones
*Your bone marrow is the factory that makes all of your blood cells and builds your immune system. It is home to thousands and thousands of **stem cells**. It is soft and spongy and protected by the hard outside of your bones.*

Bone Marrow Biopsy (Aspirate)— Procedure that collects bone marrow from the hip to assess the overall health of the marrow
Since the blood cells first start out in the bone marrow, sometimes doctors need to look at the marrow itself to really check on the health of your blood. For a bone marrow biopsy, a specially trained healthcare provider will use a special needle to collect some of the marrow from inside your bones (most commonly your hip). The procedure itself is very quick and lasts only a few minutes. You will be given medication to reduce pain, and you may be able to take medication, so you are fully asleep for the procedure. The next day you may be a little bit sore at the site of the biopsy, but most people can return to normal activity shortly after the procedure.

C

Cancer— Disease in which abnormal cells divide uncontrollably, destroy body tissue, and can metastasize; treatment includes chemotherapy and radiation
Some cells, the building blocks of our body, stop working properly. These bad cells keep growing and forming more teams of bad cells that can spread in our body and cause sickness.

Cell— Basic structural, functional, and biological unit of all known organisms
Super tiny parts of the body that fit together like LEGO blocks to build us. We have cells in our skin, our bones, our hair…everywhere. Our cells grow and renew all the time. Cancer happens when we grow too many cells too quickly.

Chemotherapy— Treatment that destroys cancer cells with drugs

Chromosome – Thread-like structures of DNA in nearly every cell that carry genes.
Big bundles of DNA that help to organize our genes. There are 46 chromosomes in total, and they come in 23 pairs. We get one copy of each chromosome from each of our parents and they are the unit through which genes get passed on.

CRISPR-Cas9 (clustered regularly interspaced short palindromic repeats)**—** Technology to selectively detect and edit certain portions of DNA
CRISPR is a very powerful tool that scientists are just starting to use to correct genetic variants. CRISPR works sort of like using the CTRL-F function in a word document. CRISPR can track through the millions of letters in the genetic code to find a very precise location for a genetic variant. Once the CRISPR has found that variant, another protein (cas-9) can come in and repair the genetic variant. While this technology gives us the potential to repair genetic diseases like RUNX1-FPD, it is also very new and scientists have to do a lot of research to make sure that there aren't side effects to using CRISPR in a specific disease or cell type. There are also challenges in designing CRISPR therapies and in ensuring that it can get into the affected cell. It may be many years before CRSIPR is able to be used to correct a disease like RUNX1-FPD, but it certainly gives us hope.

D

De novo— Variant that is not inherited from either parent
Literally means "of new" and refers to a genetic variant that happened in a person for the very first time. De novo variants are not inherited from either parent. The variants happen just like typos in the early stages of development when the DNA and genes are being copied over from the parents. The average person has ~70 de novo variants that will not be found in either parent (though most of these do not lead to disease). Once a person has a de novo variant they will be able to pass it on to their children.

DNA (deoxyribonucleic acid)**—** Molecule that carries genetic instructions in living things
DNA is the material that makes up all our genes. DNA is really just a code; we think of DNA as being written out in letters (ATC and G) and, just like any code, there can sometimes be typos or spelling errors. When we do genetic testing, we are basically just running spell-check on the genes to look for these typos.

F

Familial— Medical conditions that are shared with other family members through heredity

G

Gene— Segment of DNA and unit of heredity that codes for RNA or protein
Our genes are like our body's instruction manual or cookbook that tells us everything we need to function and grow. They get turned on at different points in our life and they make proteins. Each gene codes for a unique protein that can have diverse functions. The RUNX1 gene for example plays an important role in helping our platelets develop.

Genome— The entire set of genes in our body
If genes are like instruction manuals or cookbooks, genomes are like the entire library!

Genetic Condition— Conditions due to having an altered copy of a gene

Genetic Testing/Sequencing— Analyzing cells for changes in genes that may be a sign of a condition (such as RUNX1)
Your genes are made up of A LOT of letters, just like words in a book. Genetic testing is like running a spell-check to look for typos that can tell us if you might have a particular disease. Genetic counselors are healthcare professionals trained to order, interpret and help individuals understand the results. There are several types of genetic testing (see table below). For more information on genetic testing, visit https://www.genome.gov/About-Genomics/Introduction-to-Genomics#four.

Exome Sequencing	Sequence all protein-coding genes in your body
Next Generation/Panel Sequencing	Examine lots of genes related to bleeding or bruising (hematology)
Single Gene/Sanger Sequencing	Sequence a single gene to identify any variations in the code (applicable for single-gene disorders)

Genetic Variance (Mutations)— DNA changes that can lead to altered expression of a gene; variants may be benign, pathogenic, or of unknown significance
"Spelling changes" in DNA that lead genes, and their proteins, to be different. Sometimes spelling changes are beneficial and help create new "words" (characteristics). Other times, if your body can't read the word correctly, it can lead to diseases like RUNX1.

Germline Genetic Variant - A genetic variant that is present in nearly every cell in the body and that can be passed on to family members.
They are changes that can be inherited from parents or can be **de nov***o in the very early stages of development. Because they are present in the sperm and egg cells they can be passed on to children. They may also be referred to as constitutional variants.*

H

Hematopoietic Stem Cells— Cells in bone marrow that develop into different kinds of blood cells (eg. white blood cells, red blood cells, and platelets)
These are baby cells that start out their life in the bone marrow factory. As they grow up, they'll "shape-shift" into your red blood cells, white blood cells, and platelets. As the cells grow, they'll move into the body's bloodstream. New stem cells keep forming that can become different types of blood cells needed.

Hematopoietic Stem Cell Transplant (HSCT) or **Bone Marrow Transplant (BMT)-** Procedure wherein healthy bone marrow cells (stem cells) are transplanted into a person who has diseased or damaged marrow
Transplant just means to move. A stem cell transplant takes healthy bone marrow from one person to help replace another person's damaged cells that aren't doing their job correctly. Because we are transplanting the whole cell-making factory, someone with a bone marrow transplant can keep making new cells as they need them.

Heredity - Passing on characteristics coded for by DNA from parent to child.
We share lots of different characteristics with our family members like the way we look. These characteristics are coded for in our genes and are passed down through generations. While we can share lots of cool things with our family members, we may also share not-so-cool things like genetic diseases (such as RUNX1). This phenomenon is called heredity, and a trait is heritable when it can be passed on to family members.

L

Leukemia— A cancer of white blood cells that affects the blood and bone marrow and hinders ability to fight infection; for acute cases, treatment includes chemotherapy, sometimes followed by radiation and stem-cell transplant
White blood cells are like soldiers that battle invading infections. When you have Leukemia, your bone marrow creates white blood cells with changes that make them like injured soldiers who can't fight the infections and protect the body properly anymore.

M

Megakaryocytes— Type of hematopoietic cell
A specific type of early blood cell that will go on to produce platelets.

N

Neurons— Communication systems of nerve cells that transmit brain signals, process sensory reactions, and coordinate movement
These are cells that work as your body's telephone system. Sensory neurons tell your brain about the things you feel all the way down to your fingers and toes, and motor neurons help your brain tell the rest of your body to do things like speak or move.

P

Pathogenic— Disease-causing

Plasma— Helps blood cells and other groups (like sugars, proteins, and nutrients) reach destinations throughout the body

Platelets— Blood cells that induce clotting and stop bleeding
These are cells in your blood that act like teeny tiny band-aids. Just like when we put a band-aid on a cut to stop the bleeding on the outside, your platelets stick together in your blood so your body can stop the bleeding from the inside.

Protein— Complex molecules with diverse functions, including cell metabolism and structure of tissues and organs; composed of one or more folded polypeptide chains
"Protein" is just the word scientists use to describe the material that forms most of your body. There are many different proteins that each have a unique function - make our muscles move, our hair grow, our eyes a certain color and so on.

R

Radiation Therapy— Treatment that destroys cancer cells with X-ray radiation
Powerful x-rays will be aimed at specific parts of your body like light sabers to help get rid of cancer cells. Radiation therapy is a lot like getting an x-ray.

Red Blood Cells- Make up 44% of blood cells and provide its characteristic color; transport oxygen around the body.

***RUNX1* (the gene)**- Produces blood cells from stem cells and helps develop neurons
A gene that is the recipe for making our blood cells work correctly and helping our platelets grow.

RUNX1 (the condition)— Hereditary condition caused by an error in the RUNX1 gene; symptoms include increased bruising or bleeding. You may also see this written as **RUNX1-Familial Platelet Disorder (RUNX1-FPD).**
A condition that is caused by a change in one copy of the RUNX1 gene. It is the reason why you (and maybe some people in your family) bruise and bleed more than usual.

S

Somatic Genetic Variant – Genetic variants that happen after conception
Somatic genetic variants are genetic variants that happen as we age and are present in just a few cells in the body. Somatic variants are very normal, but if a person is found to have a lot of somatic variants all at once it can be an early sign that a cancer is forming. Your doctors may analyze your blood or bone marrow for somatic variants to help monitor you. Somatic variants are typically not found in the sperm or egg cells and cannot be passed on to family members or inherited. You may also hear somatic variants be referred to as acquired. They are the opposite of germline or constitutional variants, which are inherited and present in nearly all of the cells of the body.

T

Thrombocytopenia— Low platelet count
This is when you have too few platelets. It can take a little bit longer for your body to form its internal band-aid, and you may bleed for a little bit longer than other people.

Transcription Factor— Proteins involved in converting, or transcribing, DNA into RNA, which can then be translated into proteins. They regulate when and how much protein is made from a gene at a given time.
Transcription factors are like the genome's light switches. Your body doesn't need to make every protein all the time, so transcription factors bind to the DNA to turn genes on or off. If the transcription factor doesn't work correctly (perhaps because of a pathogenic genetic variant) it can lead to too much or too little protein being produced. RUNX1 is a transcription factor that controls genes needed for blood development.

V

Variable Expressivity— Phenomenon where different types and severities of symptoms can occur among individuals with the same genetic condition
For those with RUNX1-FPD, some may have more severe bleeding and bruising than others, and only some people will go on to develop blood cancer or leukemia. Even within families where everyone has the exact same RUNX1 variant, people can have very different experiences with the disease.

W

White Blood Cells (eosinophils, basophils, monocytes, neutrophils, T cells, B cells, and more) - Fight against infections and build immunity

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