



VHL Handbook Kids' Edition

A Handbook for
Parents and Kids
Living with
von Hippel-Lindau



VHL HANDBOOK KIDS' EDITION

A HANDBOOK FOR PARENTS AND KIDS LIVING WITH VON HIPPEL-LINDAU (VHL)

Written by Melissa Kruger,
Gayun Chan-Smutko, Christina Doyle,
and Alison Eckerman

Foreword by Anna Muriel, M.D., M.P.H.

Illustrated by Alex, Amy, Aubrey,
Carissa, James, Justin, Keri, Megan,
Mickey, and Noah

Published in the United States of America
Printed in the United States of America
First printing 2009

ISBN 978-1-929539-02-4

VHL Family Alliance

2001 Beacon Street, Suite 208
Boston, MA 02135

All rights reserved
www.vhl.org

Copyright © 2009, VHL Family Alliance
This book may not be reproduced in whole or in part
by any means (with the exceptions of short quotes for
the purpose of review), without written permission of
the VHL Family Alliance

TABLE OF CONTENTS

FOREWORD: A Note To Parents
By Anna Muriel, MD, MPH

CHAPTERS:	Page
1. INTRODUCTION	6
2. WHAT IS VHL?	7
3. HOW DO PEOPLE GET VHL?	10
4. WHAT ARE DNA AND GENES?	11
5. WHAT HAPPENS IF I HAVE VHL?	15
6. WHAT IF MY VHL SYMPTOMS CAUSE A PROBLEM?	18
7. WHAT CAN I DO TO STAY HEALTHY?	20
8. HOW WILL VHL CHANGE MY LIFE?	21
9. SHOULD I TELL PEOPLE THAT I HAVE VHL?	22
10. CAN I TALK TO OTHER KIDS WHO HAVE VHL?	23
11. WILL THERE EVER BE A CURE FOR VHL?	24
WORD LIST GLOSSARY	25
ABOUT THE AUTHORS	29
ABOUT THE ILLUSTRATORS	30
REFERENCES	30
ACKNOWLEDGMENTS	30

Dedicated to the health and
happiness of all children
and families.

FOREWORD: A NOTE TO PARENTS

This handbook is written specifically as a guide to help you talk about von Hippel-Lindau disease with your children. It is meant to give children of all ages a basic idea about VHL. It can also be used as a starting point for discussions about how VHL has affected your family. The book may be most helpful when a member of the family has been directly affected, and your children may be facing testing themselves.

You know your children best. It is suggested that you read the whole book first on your own, and then think about how it could be useful to them. Your approach may depend on their ages and how they prefer to take in information. Determine whether your children would like for you to read it aloud to them and discuss it, or if they would like to read it themselves and then have a discussion. You may find that your children do not want to read it at all. However, you can still get ideas from the book about how to explain VHL in terms that they will understand.

You may begin the discussion by asking your children what they already know about VHL, if anything, and encouraging them to ask questions that they already have. Try to clarify the questions to understand any underlying concerns, and be open to your children's questions. Their questions may come immediately after receiving the information or as they process it over time. As with any information that you or your children receive, be it from your health care providers, other families affected by VHL, the Internet, or other sources, be sure that you take the time to sort out misconceptions, and find out what is specifically relevant for you and your family.

We hope that this book will be helpful as you cope with VHL.

Anna Muriel, MD, MPH
Staff Child Psychiatrist, Massachusetts General Hospital
Co-author, *Raising an Emotionally Healthy Child when a Parent is Sick*

Parents will also need a copy of
The VHL Handbook: What You Need to Know about VHL
which includes information to share with your doctor about preventive screening for people of all ages.

CHAPTER 1: INTRODUCTION

What is this book about?

The **VHL Handbook Kids' Edition** was written for kids of all ages who live all over the world. This book will help you and your parents learn about **von Hippel-Lindau** or **VHL**. Perhaps you or a loved one has VHL. The book will explain in simple terms what VHL is, how people get it, and how people live with it.

Read this book with your parents so they can explain things to you and answer any questions that you may have. If you like, you and your parents can read one chapter at a time in order, or just read any chapter of your choice. Do not feel like you have to read the whole book at one time because there is a lot of information to be learned. You may also decide to read certain chapters again at a later point in time. However you decide to use this book is up to you and your parents.

You will find a lot of new words that are typed in **bold** print. We explain these new words at the end of the book in the Word List Glossary. You will also find many questions that kids with VHL might ask. These questions are underlined. The answers follow the questions in the paragraphs directly below them.

We hope that this book will help you better understand VHL. This book is meant to leave you with a positive feeling about VHL so that you can continue to live your life to its fullest!



Illustration by
Keri K., Age 6

CHAPTER 2: WHAT IS VHL?

What is VHL?

VHL stands for **von Hippel-Lindau** disease. It is named after two doctors who first discovered it: Dr. Eugen von Hippel and Dr. Arvid Lindau. VHL is a rare disease, which means that compared to other known diseases, there aren't many people in the world who have it. But, even though it is rare, there still are thousands of people around the world who do.

Just because a person has VHL, it does not mean they are sick. Most people with VHL usually feel fine. Having VHL means that a person has a greater chance of growing tumors or cysts in certain parts of their body than someone who doesn't have VHL.

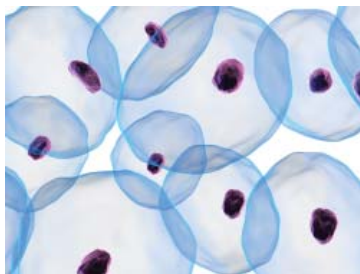
A Cell



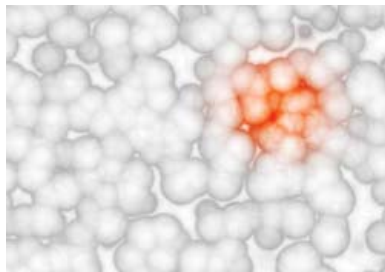
What are tumors?

Our body is made up of millions of **cells**. Each cell has a job to do, such as being a skin cell, a brain cell, or a kidney cell. Healthy cells normally make copies of themselves as they grow at a normal pace. But, if a kidney cell starts growing faster than normal, a tumor can form in the kidney. A **tumor** is a clump of cells that do not behave like normal cells anymore. With people who have VHL, their tumors grow like small knots bunched together.

A Group Of Cells



Cells Turning Into A Tumor



People with VHL can also grow **cysts**, which are not tumors. A cyst is a collection of fluid or liquid that is often found in the **kidneys** and **pancreas**. Most cysts in the abdomen do not cause health problems, and the organs usually work normally with them.

What kinds of tumors can someone with VHL get?

People who have VHL may have tumors in different parts of their body. These tumors could be found in the following areas:

Brain: Your brain is located inside your head; it is your body's main control center for your nervous system; your brain helps you think; and it also controls your how your body works.

Spinal cord: Your **spine** is a set of bones, like a stack of rings, that run down your back. It is also called your backbone. The green line in the drawing represents the **spinal cord**, running like a wire down through those bones, carrying signals between your brain and all parts of your body.

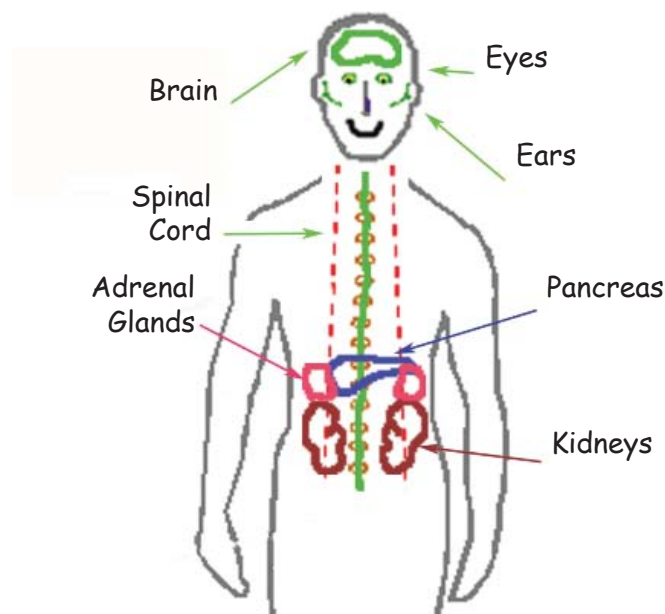
Eyes: Your eyes are organs of sight, which allow you to see and detect light.

Ears: Your ears are organs of hearing, which allow you to listen to sounds; your ears are also important for your sense of balance.

Kidneys: Your two kidneys are located in your abdomen or stomach area; your kidneys are organs that filter your blood and get rid of waste products by turning them into urine.

Adrenal glands: Your adrenal glands are found above your kidneys; they control hormones or chemical messengers that send signals from one cell to another.

Pancreas: Your pancreas is an organ that is behind your stomach; it helps digest the food in your stomach.



Do all people with VHL grow tumors?

Every person is different, even within the same family.

- Some people may have a few tumors during their whole life, some people may have more, and some people may never have any.
- Some people may only have a tumor in one part of their body, and some people may have tumors in different parts of their body.
- Some people may even have tumors grow in one part of their body more than one time.

If you get a **tumor** it doesn't mean that you need to be scared. Your doctors will spend most of their time watching it closely in order to make sure it doesn't cause a problem.

People with **VHL** are not the only people who can get tumors. Anyone can get a tumor anywhere in their body whether they have VHL or not.

Illustration by
Amy A., Age 10



Do people with VHL get cancer?

Tumors sometimes turn into cancer and sometimes they do not. **Cancer** happens when certain types of tumors grow too much and spread to places in the body they shouldn't. But, most of the time, VHL tumors are not cancerous. Sometimes tumors that grow on the **kidneys** can turn into cancer, but if doctors catch these tumors early, they can remove them if needed. Anyone can get cancer, not just people with VHL.

CHAPTER 3: HOW DO PEOPLE GET VHL?

How do people get VHL?

VHL is a **hereditary** disease, which means that it is found in families. It can be passed on from your grandparents to your parents and then to you. Most likely one of your parents also has VHL. A parent with VHL can pass it on to you just like other traits (hair color or eye color) without knowing it. You may have other family members who have VHL as well, like a grandparent, brother, sister, aunt, or uncle. VHL is not the only disease found in families. There are many other diseases that are hereditary as well.

You cannot get VHL from another person by touching them or by catching their cold. Every person who has VHL is born with it.



Illustration by Carissa K., Age 9

In very rare cases, VHL may not be passed on from a parent to a child. Sometimes a person is the first one in their family to have VHL. This does not mean that they did anything that caused them to have VHL; they were just born that way.

How do I know if I have VHL?

You may have already had symptoms related to VHL and were checked by your doctor. A **symptom** is a feeling, sensation, or pain that usually makes you feel different than how you normally feel every day. It's also possible that you do not have any symptoms, but you have a family member who has VHL. In both cases, you and your parents may want to test for VHL so that your doctors can make sure that you are taken care of.

The only way to truly know if you have VHL is to have **genetic testing** done. Genetic testing is explained in Chapter 4. You and your parents, doctors, and **genetic counselor** will decide if this test is right for you.

CHAPTER 4: WHAT ARE DNA AND GENES?

In order to understand how **genetic testing** works, and since **VHL** is a **hereditary** disease, it is important to learn about DNA and genes. This is how we inherit or get traits like our hair color, eye color, or even diseases from our parents.

What is DNA?

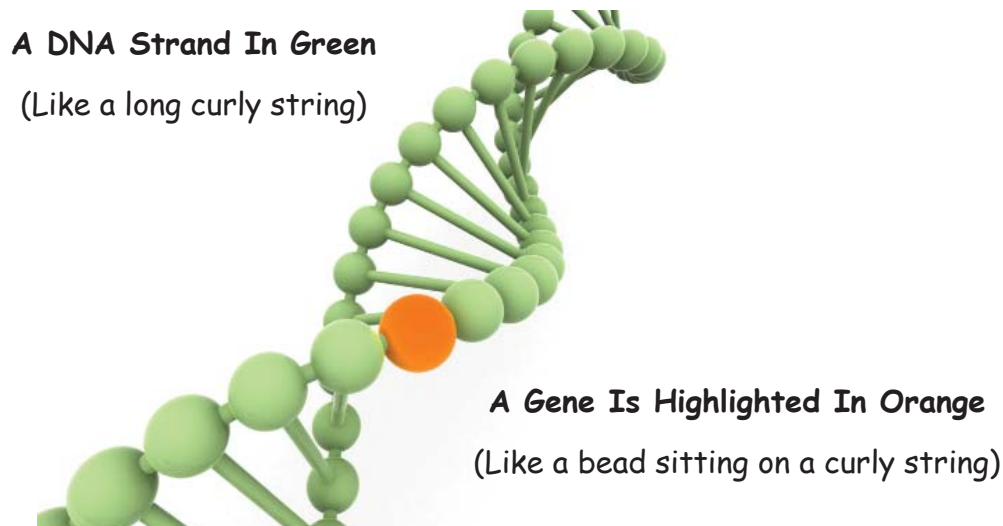
DNA stands for deoxyribonucleic acid, which is too hard to say. To make it easier, we just call it DNA. It is found in every cell of your body. The DNA is very tiny, and you are unable to see it, but it is inside of you. Under a microscope it looks like long curly strings.

What are genes?

Your **genes** are chunks of DNA, sitting like beads on the long curly strings. Genes are a special code that carry the directions for how your cells should work. Genes are the instructions your body follows on how to create you! You have all kinds of different genes in your body. Some of your genes will tell your cells how to be skin cells or heart cells. Each gene and cell has a special job to do.

You were born with two copies of every gene in your body. One copy came from your mother, and one copy came from your father. It is estimated that a person has 25,000 to 35,000 genes in their body!

You may have a gene for brown hair from your mother and a gene for curly hair from your father. Maybe you have a gene for green eyes from both your mother and your father. Your parents pass their genes on to you, which make you a very special person!



How do DNA and genes work in my body?

Think of the **DNA** in your cells like a library full of books. The DNA in your cells is divided into sections called genes. **Genes** are just like books in a library that are made up of words and letters. Each gene carries instructions to tell your cells how to grow, how to do their job, and how to stay healthy.

Just like a spelling mistake can happen in a book, there may be a change in the gene code within a person, called a **gene mutation**. That is what **VHL** is, a gene mutation, like a tiny spelling mistake in a book. That means the VHL book, or gene, does not make sense to the cells in your body.

DNA = A Library

Genes = Books In A Library

Gene Mutation = A Spelling Mistake In A Book

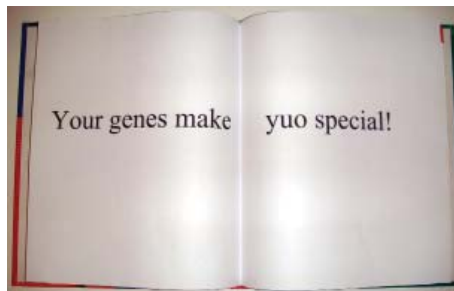
DNA = A Library



Genes = Books in a Library



Gene Mutation = A Spelling Mistake In A Book



How do people get the VHL gene mutation?

Since you get half of your **genes** from your mother and half of your genes from your father, you have two copies of every gene in every cell of your body.

People who do not have **VHL**, have two working copies of the VHL gene. They do not have a mutation in their VHL genes. But people who do have VHL, have a mutation in one copy of the VHL gene, and sometimes that gene doesn't work right.

Every time a baby is born to a parent who has VHL, there is a 50% chance that the baby will have VHL, and there is a **50%** chance that the baby will not have VHL. It doesn't matter if the baby is the first child in the family, the second, the third, or the fourth. The chance of getting the VHL **gene mutation** is the same every time a baby is born.

Thus, if you have one parent who has VHL, there are two different outcomes that you may have.

- You have VHL: You have a copy of the VHL gene that works, and a copy of the VHL gene that has a mutation.
- You do not have VHL: You have both copies of the VHL gene that work, so your parent who has the VHL gene mutation passed on their working copy to you.

If you have VHL, as long as the working copy of the VHL gene is fine, the cells stay healthy.

Parent 1: With VHL

A = VHL Gene Mutation
B = VHL Working Gene



Parent 2: Without VHL

C = VHL Working Gene
D = VHL Working Gene



Each child gets one copy of the VHL gene from each parent.
In this way, there are four possible arrangements of these four genes:

AC: Child With VHL



A = VHL Gene Mutation
C = VHL Working Gene

AD: Child With VHL



A = VHL Gene Mutation
D = VHL Working Gene

BC: Child Without VHL



B = VHL Working Gene
C = VHL Working Gene

BD: Child Without VHL



B = VHL Working Gene
D = VHL Working Gene

How is genetic testing done?

The **genetic testing** is typically done with a **blood test**, which is a test where a small sample of blood is taken from your arm. Some people think it feels like a poke on your arm. It doesn't hurt very much or for very long, especially if you relax. Genetic testing can sometimes be done with a **cheek swab** as well, which is like taking a small brush (about the size of a Q-tip) and rubbing it on the inside of your cheek to pick up some of your cheek cells. The cheek swab test does not work for all families, so your doctor will tell you which test is required to get the best information.

Your blood sample or cheek cells are examined in a **laboratory** to see if you have **VHL**. The test looks for the **VHL gene mutation** in one of your VHL copies, which is like looking for a spelling mistake in the VHL book.

There are two possible results of the genetic testing:

- If the test is positive, then you have VHL.
(One of your VHL copies has the VHL gene mutation.)
- If the test is negative, then you do not have VHL.
(You have both working copies of the VHL gene.)

What if I'm nervous about the test?

The **blood test** might seem scary. That could be because you may be learning new information about yourself even though you feel fine, or because you have **symptoms** and you are worried about them. The important thing is to talk to your parents about these feelings. It might be hard to wait for the **blood test** results. Waiting is always difficult. You may have to wait a few weeks. If you can, try to get your mind off of things for a while as that can help. Eventually, your doctor or your **genetic counselor** will call your parents to talk about the results. Just remember that whatever the result is, it doesn't change who you are!



Illustration by
Mickey P., Age 8

CHAPTER 5: WHAT HAPPENS IF I HAVE VHL?

What happens if I have VHL?

If you have **VHL**, your doctor will do a physical exam to make sure you stay healthy. The physical exam usually happens every year. There are a set of guidelines that help your doctor figure out which tests are best for you depending on your age and your family background. Most of these tests will be done just once a year too.

The tests that your doctor orders are to monitor what is going on inside of your body. To monitor something means to keep a close watch on it, and your doctor does this by running tests. We call these tests **screenings**. These tests usually do not hurt, but you might feel a little uncomfortable with some of them because they are strange or new to you. Your doctor will be able to explain the tests to you before you have them done.

What kind of tests or screenings would I need?

Eye Exam: Most kids need to have their eyes checked regularly whether they have VHL or not. But, if you have VHL, you would need to see an **ophthalmologist** or a **retinal** specialist who is an eye doctor. Eye doctors can look through your eyes to see what is going on behind them. Sometimes tiny **tumors** can hide out there, so your doctor will use a special magnifying lens called an **ophthalmoscope** to look at your eyes very closely.

Eye Exam



The eye exam does not hurt. Basically, your eye doctor will shine a light into your eyes and ask you to look around in different directions. Before the exam, your doctor will put a few drops into your eyes to dilate them. The drops make your pupils (the small black center of your eyes) get slightly bigger. This will help your doctor to see your eyes better. You should bring sunglasses with you because your eyes might be sensitive to light afterwards, but this soon goes away.

Hearing Exam: You will also need a hearing exam by an **audiologist**. This is a fancy name for an ear doctor. It is like the hearing tests given at schools; only it will last just a little longer. You usually have to wear earphones and listen for beeps. The test doesn't hurt at all. Some kids think it is fun to listen for the beeps!

Hearing Exam



Urine/Blood: Another test you will need to take is a **24-hour urine test** or a **blood test**. These tests will be able to tell your doctor about possible **tumors** called **pheochromocytomas** or pheos that you might have on or near your **adrenal glands**. If you do the 24-hour urine test, you basically have to pee in a cup for a whole day and pour it into a big jug each time, which you keep in your refrigerator. You will need to take the jug to a **laboratory** to be examined. During this test, it is a good idea to stay home so you don't forget to collect your urine.



If your doctor wants you to have the blood test done instead of the urine test, you will probably just go to the laboratory. Sometimes your doctor will tell you not to eat for a few hours before taking the test. You may want to have the test done in the morning before breakfast so that you aren't too hungry.

Abdominal Ultrasound: **VHL** patients need to have an **ultrasound** exam too. For this test, you lay on your back, and your doctor will put some warm clear jelly (not the kind you eat) on your stomach. Then your doctor will rub an instrument similar to a computer mouse across your stomach. While your doctor does this, he or she looks at a computer screen to see what your insides (or organs in your abdomen) look like. It doesn't hurt at all, in fact, it kind of tickles!

Abdominal Ultrasound Exam



MRI - Abdomen, Brain, Spine: There are also some special x-ray machines that can see just about everything in your body from head to toe. One of these machines is called an **MRI**. The MRI is big machine that you lay down in. It is very loud and noisy, but you won't feel anything. It is important to be very still when you have this test so the MRI machine can take clear pictures. Your doctor will usually give you ear plugs so you will not hear the loud noises. Some places with MRI machines even let you listen to your favorite music CD during the test!

Sometimes in the middle of the test, your doctor may give you a small injection or shot of dye to highlight certain parts of your body on the pictures that the MRI machine is taking. For the most part, it is an easy test. Some people rest or take a little nap. You can even bring your teddy bear with you to hug.



MRI Exam

CHAPTER 6: WHAT IF MY VHL SYMPTOMS CAUSE A PROBLEM?

What if my VHL symptoms cause a problem?

Most of the time a person with **VHL** feels fine, but you will need to have regular **screenings**, like we talked about in Chapter 5. Even if a **tumor** is found, it may not need to be taken out, and your doctor will keep a close watch on it.

Sometimes a tumor causes a person to have **symptoms** that they never had before. If you have a symptom that is caused by a tumor and does not go away, your doctor might say that you need to meet with a different doctor who knows how to take out or remove **tumors**. This special doctor may be a **surgeon**.

It is important to know that even if you have a tumor removed, you may have another tumor grow in the same place at another point in time. That sometimes happens with people who have VHL. But, as long as you keep up with your screenings, your doctor should be able to catch things early.

How are tumors treated or removed?

There are different ways to treat tumors. Your doctor and your parents will tell you more about the kind of treatment you might have if you ever need it. Different treatments are used for different types of tumors in the body. The area of the tumor will also determine what type of treatment you may have. For example, eye tumors can often be treated with a tiny laser aimed only at the tumor cells. Your doctors will also talk to you about what you can expect during the treatment and after.

All Better After Surgery



Sometimes doctors or surgeons use **surgery** to remove tumors. There are many different types of surgeries. Another word for surgery is an **operation**. Usually your doctor will give you a special medicine to make you fall asleep for a short time. During this time, your doctor will take out your tumor, but because you are asleep you will not feel anything. It does take time to feel completely better after a surgery, so you will need to rest and follow your doctor's directions. During your recovery, it may be a good time to read a book or watch your favorite movie at home. After your tumor is removed you should feel better and back to normal very soon.

What symptoms should I look out for?

It is important to tell your parents if you do not feel well or if you feel unusual at any time even if it is something that isn't that bothersome. For example, if you are getting headaches more often or if your vision changes at all, then tell your parents right away. If you are not with your parents, and you are feeling sick, be sure to tell the person responsible for you that you have **VHL** or that it runs in your family.

Maybe you have a **symptom** that doesn't seem important, maybe you think it will go away, maybe you want to ignore it, or maybe you are too embarrassed to talk about it. A symptom you are feeling could be a clue to what is going on inside of your body.

Just because you have a symptom does not mean that you have a **tumor** or that you need to be scared. The symptom or feeling may be related to VHL or it may not be. Your parents will know how to best help you, and they will know when you need to see your doctor. It's very important for everyone to pay attention to their bodies, not just people with VHL.

Here is a list of symptoms that you should look out for:

- Headaches: If your head hurts and doesn't feel right at any time.
- Vision Problems: If your eyes are blurry, if you see spots, or if your vision doesn't seem normal at any time.
- Hearing Problems: If your hearing changes, if you are hearing less out of one ear than the other, or if your hearing doesn't seem normal at any time.
- Vomiting: If you throw up or feel sick to your stomach at any time.
- Balance Problems: If you feel like you can't stand up straight, or if you feel dizzy at any time.
- Pain: If any part of your body hurts at any time.
- Not Feeling Normal: If you do not have energy, feel tired, if your heart beats really fast, if you are often sweaty, if you feel nervous, or just feel sick at any time.

Illustration by
James B., Age 12



CHAPTER 7: WHAT CAN I DO TO STAY HEALTHY?

Can I still be healthy and have VHL?

Yes! Most people who have **VHL** live healthy and happy lives. The number one thing you can do to be in control of VHL is to visit your doctor every year for a physical exam and to have your **screenings** done on time. If you and your doctor are on top of your screenings, you should be able to catch any problems early that might come up. This will make it easier for your doctor to monitor you and to take care of any **symptoms** that you may have. Just remember to tell your parents if you do not feel well at any time so they can help you.



Illustration by Alex A., Age 12

What else can I do to stay healthy?

Use your healthy habits. It is important for everyone, not just people with VHL, to eat healthy foods, exercise (if permitted by your doctor), and to get enough sleep. Eat plenty of fruits and vegetables and keep up a balanced diet. If you keep your body as healthy as possible, it will be easier for you to fight off any problems with VHL if they should ever come up.

Is there anything else I can do?

Keep a positive attitude! Keeping your mind healthy and happy is just as important as keeping your body healthy and happy. If you are ever feeling sad or unsure about anything, talk about it with your parents, family members, teachers, doctors, friends, or a **counselor**. A counselor is a special person who talks to people of all ages to help them feel better inside. Talking about any type of problem with someone can make you feel better. People are here to help you!

CHAPTER 8: HOW WILL VHL CHANGE MY LIFE?

Will VHL change my life?

All kids have different feelings about having **VHL**. Your everyday life may not really change, and you will still be the same person you are, but the way you think about things may change. You may have different feelings about VHL at different times. There may be times that you will not even think about it, and there may be times that you will.

Illustration by
Noah S., Age 11



You might feel:

- Like you are the only kid that has VHL.
- Mad and upset that you have VHL.
- Sad that you have VHL.
- Like life isn't fair.
- Afraid that VHL might make you sick in the future.
- Scared about having tests or **surgery**.
- Tired of doing tests and visiting doctors.
- That VHL is a really big problem and is too much to handle.
- That VHL is not really a big deal.
- Confident that your doctors know what is happening inside your body.
- Glad that your parents and doctors are helping you to stay healthy.
- Happy that you are still the same special person you always have been.

You may also have other feelings that are not on this list. No matter how you feel now, it's okay. All of your feelings are normal, and it's okay to have them. VHL may seem very scary, but it does not have to be. You are not alone, and you are not the only kid who has VHL. Most kids have happy and healthy lives, even though they have it. It may help to talk to other kids who have VHL, and Chapter 10 will give you information on how to communicate with them.

CHAPTER 9: SHOULD I TELL PEOPLE THAT I HAVE VHL?

Should I tell people that I have VHL?

People with **VHL** do not look any differently than those without VHL. No one will be able to look at you and know that you have it. The only way people will know you have VHL is if you decide to tell them. You may feel that you don't want anyone to know, and that's okay. You may decide to tell a few people only, and you may decide not to tell anyone. It is all up to you.

Right after you find out that you have VHL, you may not feel like talking about it. It may be hard to talk about, even with people whom you are very close to. That's okay and completely normal. If you do talk about it eventually, you will feel better. First, talk to your parents or someone you are very close to. Later, you may decide to tell your friends. Your parents can help you decide which friends to tell, how to tell them, and when to tell them. Perhaps you would only like to tell your close friends, the kids you trust, and the kids who care about you.

Will my friends treat me differently because I have VHL?

A true friend will not treat you differently because you have VHL. True friends will know that you are still the same person you always have been. VHL does not define who you are, and it does not make you different. You will always be a special person whether you have VHL or not.



Illustration by
Aubrey W., Age 7

CHAPTER 10: CAN I TALK TO OTHER KIDS WHO HAVE VHL?

Where can I find kids who have VHL?

Since **VHL** is rare, it may seem hard to find other kids with VHL, but believe it or not, there are kids all over the world who have it! A great way to find and communicate with other kids who have VHL is through the Internet (with your parents' permission).

If you visit the vhl.org website, you can find links to support groups for kids of all ages. You can start a chat group, read comments, make friends, etc. Just like you, other kids with VHL understand the feelings you have, and it would be helpful to communicate with them. You never know, you may meet lots of new friends!

Who else can I talk to for help?

If you ever need to talk to anyone, don't be afraid to talk to your parents. Your parents can even make an appointment for you to talk to a **counselor**. As we mentioned in Chapter 7, counselors understand people's feelings very well, and it is their job to help you out in any way they can.

Remember, there is always someone who is there for you. If you or your family ever need more advice or support, you can always contact the VHL Family Alliance at (800) 767-4VHL or visit www.vhl.org.



Illustration by
Justin S., Age 7

CHAPTER 11: WILL THERE EVER BE A CURE FOR VHL?

A Cure for VHL?

Right now in 2009, there isn't a cure for **VHL** yet. However, there may be a pill someday that a person can take to slow down or stop **tumors**, maybe even in our lifetime!

Scientists and **researchers** are studying VHL and other diseases to help prevent them or control them. Medical science is learning more and more about **DNA** and **gene therapy**. It may be possible that one day doctors could replace or fix a mutated VHL **gene** with a normal, working gene.

Is there anything I can do to help find a cure for VHL?

Yes! You can help raise awareness of VHL which means telling a lot of people what VHL is. Since it is rare, there are many people that have never even heard of VHL. You can also create, contribute to, or help fundraisers for the VHLFA - von Hippel-Lindau Family Alliance. Encourage your family members who have VHL to join an available **clinical trial** so doctors and researchers can learn more about new treatments.

Can kids my age help too?

Yes! Kids your age may be the ones to find a cure for VHL! So keep up your studies in school and encourage your friends to remember that education is very important, not only to them, but to everyone. The more we raise awareness together and the more we learn about VHL together, the faster we can find a cure together!



Illustration by
Megan K., Age 6

WORD LIST GLOSSARY

50% (fifty percent): A 50% chance is one chance out of two possible outcomes, like a coin toss. If you flip a coin, there are only two ways it could land: heads or tails. Each time you flip a coin, there is a 50% chance of it landing with heads up.

Adrenal Glands: These glands are located on top of each kidney (most people have two). They help control hormones, or chemical messengers, that send signals from one cell to another. People with VHL may get tumors called pheochromocytomas in the adrenal glands.

Audiologist: An ear doctor who gives hearing exams (audiograms) to determine hearing loss and function.

Blood Test: A test where a doctor or a nurse will take a sample of a person's blood using a needle. The test doesn't hurt much if you relax, and some people say it feels like a poke on the arm.

Brain: The brain is located in the head; it is the body's main control center for the nervous system. The brain controls bodily functions. It also controls thought and reason. People with VHL may develop tumors in the brain.

Cancer: Cancer happens when healthy cells do not grow in an orderly way. Cancerous tumors are called malignant tumors. Malignant tumors can destroy healthy cells that are near the tumor and possibly spread to other parts of the body.

Cells: A cell is the smallest unit, or building block, of all people and other living things. Each cell has a special job to do like being a brain cell or kidney cell. Every cell in a person's body contains DNA and genes, which have a set of instructions to tell the cells what to do. A person is estimated to have 100 trillion cells in their body.

Cheek Swab: A cheek swab is taken with a small brush by rubbing it on the inside of a person's cheek in order to pick up cheek cells that contain DNA. The DNA is used for genetic testing.

Clinical Trial: A clinical trial is the study of people with certain diseases by doctors and researchers. Doctors perform different tests or give different medications to people in a clinical trial in order to see if the tests or medications make their health better. This is how doctors and researchers can potentially find cures or medications to prevent and control diseases.

Counselor: A professional person (psychological expert) who helps people of all ages deal and cope with disease, difficulties in life, stress, etc. A counselor is a person who talks to people alone or with their families based on their situations and needs.

Cyst: A cyst is a collection of fluid or liquid. Cysts in the abdomen usually do not cause symptoms or physical problems. Organs usually function normally with them.

DNA: DNA stands for deoxyribonucleic acid. DNA is found in every cell of the human body. DNA contains genetic instructions called genes which tell the cells in a person's body how to work. Every person gets half of their DNA from their mother and half of the their DNA from their father.

Ears: The ears are organs of hearing, which allow a person to listen to sounds. The ears also play a role in a person's balance. People with VHL may get tumors in the inner ear.

Eyes: The eyes are organs of sight, which allow a person to see and detect light. People with VHL may get tumors in the retina (part of the eye).

Gene Mutation: A change in the instructions or sequence of a gene; there are many different mutations that can occur. Sometimes a mutation is never discovered, sometimes a mutation may cause cells to not behave normally, and sometimes a mutation may cause a disease in a person, like VHL.

Gene Therapy: Gene therapy is a technology that is still being explored and learned about by doctors, scientists, and researchers. The idea of gene therapy is to replace or repair genes that are not working well with healthy working genes into a person's cells.

Genes: Genes are found in every cell of the human body on long curly strings of DNA. Genes carry the directions on how cells should work. Genes are the blueprints for how to create a person. Every person has two copies of every gene in their body, one they got from their mother, and one they got from their father. It is estimated that a person has 25,000 to 35,000 genes in their body.

Genetic Counselor: A professional person (medical genetics expert) who helps patients and families cope with genetic diseases. The genetic counselor also gives information to patients and helps them to find doctors who can treat their needs.

Genetic Testing: A test that looks at the genes within a person and finds gene mutations. The test is usually performed in a laboratory with a blood sample.

Hereditary: When genes are passed on from a parent to their child. Certain characteristics like hair color and eye color are hereditary, as well as some diseases like VHL.

Kidneys: The kidneys are located in the abdomen. Kidneys are organs (most people have two) that filter blood and get rid of waste products by turning them into urine. People with VHL may get tumors in the kidneys.

Laboratory: A laboratory, also called a lab, is where lab technicians, doctors, researchers, and scientists work. A lab is a place where research and experiments are performed. In medical labs, blood and urine may be tested in order to get health information about a patient.

MRI: MRI is an abbreviation for Magnetic Resonance Imaging. This is a machine that takes pictures of the inside of a person's body. The machine uses magnetic energy, so there is no radiation used. The pictures appear on a computer screen. The MRI machine can take images of soft tissue (like kidneys) or hard tissue (like bones).

Operation: See the definition for surgery.

Ophthalmologist: An eye doctor who specializes in diseases and surgery of the eye.

Ophthalmoscope: A magnifying lens used by an eye doctor to examine the eyes. This tool can be used to see the health of the retina.

Pancreas: The pancreas is a gland organ that is behind the stomach. It aids with the digestion of food. People with VHL may get tumors in the pancreas.

Pheochromocytomas: A tumor of the adrenal gland, which is also called a "pheo" for short. Pheos can sometimes be found in other areas outside of the adrenal glands.

Researchers: There are many different types of professional researchers. Researchers study many different things. They investigate, discover, learn, and share their knowledge with others.

Retina: The retina is the nerve tissue located at the back of the eye. It is like the film in a camera because it takes pictures that a person sees. The pictures are sent through the optic nerve to your brain for understanding. People with VHL may get tumors in the retina. A retinal specialist is an eye doctor who specializes in treating the retina.

Scientists: There are many different types of professional scientists. Scientists study many different things like the body, animals, the earth, the universe, etc. Scientists perform tests or experiments in order to learn more about their area of study.

Screenings: Tests that a doctor orders to monitor what is going on inside a person's body. In VHL patients, there is a recommended schedule for screenings based on age. Most people need to have their screenings done once a year, but this will also depend on the person and their family history.

Spine: The spine is located in the back. It is also called the backbone. The spine is made up of small bones called vertebrae that hold you up and protect the spinal cord, which contains many nerves.

Spinal cord: The spinal cord is a bundle of nerves like a bunch of wires that run from your brain down through the holes in the vertebrae of your spine. It carries signals between your brain and other parts of your body. People with VHL may get tumors in the spinal cord.

Surgeon: A doctor who performs operations. There are many different types of surgeons. Surgeons specialize in the treatment of different parts of the body.

Surgery: A way doctors can remove tumors, treat injuries, or improve how the body works. There are many different types of surgeries. In general, a surgery (operation) is done when a person is asleep for a short time at a hospital. While the person is asleep, the doctor will do the surgery. Most people need time to recover or get better from a surgery. The time needed to feel better is based on the person's health and the type of the surgery they had.

Symptom: A feeling or sensation that makes a person feel different from how they normally feel every day. Sometimes a symptom may be a pain or weakness in a certain part of the body. If a symptom does not go away, a person should be examined by a doctor in order to figure out what the cause is.

Tumors: A collection of cells that do not behave like normal cells any more. Tumors can either be benign (not cancerous) or malignant (cancerous).

Ultrasound: An ultrasound machine that uses sonar or sound by bouncing sound waves off the objects inside the body to create images or pictures of specific organs. The doctor or nurse rubs a probe with jelly over the part of the body that needs to be checked. The pictures appear on a computer screen. There is no radiation with ultrasounds.

Urine Test (24-Hour): A test used to check for levels of catecholamines and metanephrines (hormones or chemicals) found in the urine. Urine is collected for a 24-hour period in a special jug, which should be refrigerated. After the test is done, it needs to be taken to the laboratory to be examined.

von Hippel-Lindau: A rare, hereditary disease that may cause tumor growth in various parts of the body. The disease is named after two doctors who first found it: Dr. Eugen von Hippel from Germany and Dr. Arvid Lindau from Sweden.

VHL: Stands for von Hippel-Lindau disease.

ABOUT THE AUTHORS:

Gayun Chan-Smutko, MS, CGC, Co-Author



Gayun has a Master of Science degree in Genetic Counseling from Brandeis University and is certified by the American Board of Genetic Counseling. She received a Bachelor of Science in Cellular, Molecular Biology from the University of Michigan. She is a mother and a senior genetic counselor at the Massachusetts General Hospital Cancer Center. Since 2002, Gayun has enjoyed the privilege of coordinating the VHL Comprehensive Care Center.

Christina Doyle, Co-Author



Christina has a Bachelor of Arts degree in Sociology from the University of California, San Diego. She will be attending graduate school this fall, 2009. She plans to become a Genetic Counselor. She and her father have VHL.

Alison Eckerman, Co-Author



Alison is a mother of two. Her husband and her two children have VHL. She has done extensive research over many years in order to ensure that her family has dealt with this disease as effectively as possible. In doing so, Alison's experiences have been able to help others as well. She has written articles for the VHL Family Alliance and contributes to the VHL messages boards.

Melissa Kruger, Co-Author & Editor-In-Chief



Melissa has a Bachelor of Science degree in Child Development and a California Multiple-Subject Teaching Credential from California State University, Fullerton. She is a mother of two and an elementary school teacher. Melissa and her daughter both have VHL. Her father, grandmother, and aunt also have the disease.

ABOUT THE ILLUSTRATORS:

Our young illustrators are children with VHL and children who have a family member or a friend with VHL. They have all volunteered their time and imaginations to help with this book. We know a lot of love went into their precious drawings!

REFERENCES:

Genetic Alliance website. (Information and publications for professionals and patients). <http://geneticalliance.org>

National Institutes of Health website. <http://www.nih.gov>

New International Webster's Student Dictionary. (1996).

Von Hippel-Lindau Family Alliance. (2005). *The VHL Handbook: What You Need To Know About VHL* (3rd ed.). Boston, Massachusetts

Von Hippel-Lindau Family Alliance website. (Information for families, clinicians, researchers). <http://www.vhl.org>

Royalty-Free Images From:
<http://www.dreamstime.com> and <http://www.fotolia.com>.

ACKNOWLEDGMENTS:

Thank you to Anna Muriel, MD, MPH for advice on our book and for writing the informative foreword for the parents.

Our thanks to Deborah L. Jones and her family for providing partial funding for this project.

We would also like to thank all of the professional and parent reviewers for taking their time to read and provide valuable comments for the improvement of our book.

Lastly, we would like to recognize a wonderful children's guide called **FAP & Me**, which served as a model and a starting point for our book. **FAP & Me** is a publication of the National Society of Genetic Counselors.