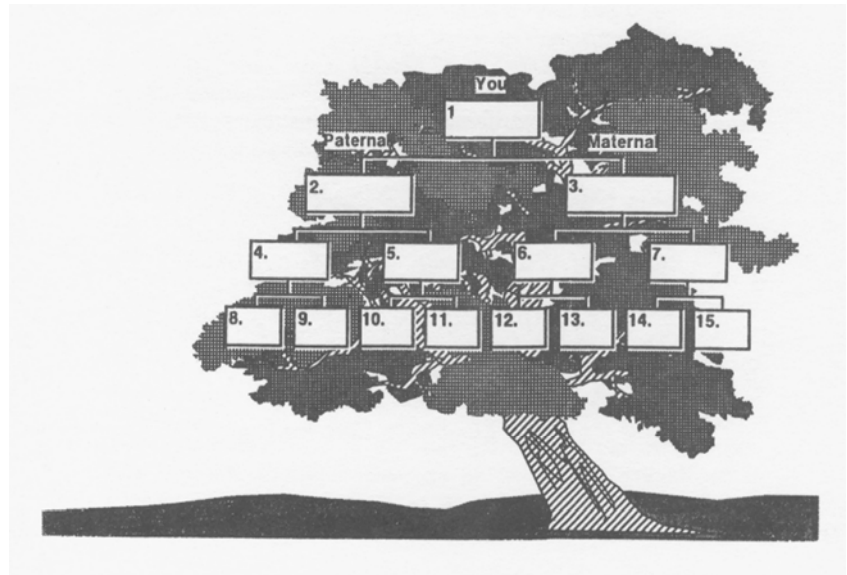


# Your Family Health Tree

by  
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*With early detection and treatment, there is more hope today for families with Von Hippel-Lindau syndrome than ever before. By compiling a Family Health Tree you can significantly assist your health professionals to locate family members at risk, diagnose the condition early, and contribute to their good health.*

If you are like most people, you probably think that genetic diseases are rare. While it is true that each of the more than 5,000 genetic disorders is relatively rare, taken as a group they are quite common. Innumerable people suffer from disorders due wholly or in part to defective genes or chromosomes. Genetic diseases are far more prevalent than is generally realized, as the statistics below indicate:

- 12 to 15 million Americans have a genetic disorder of one kind or another.
- 20 million Americans are carriers of true genetic defects
- One out of every 250 newborn babies has a genetic disorder.
- One out of every three babies or young children admitted to a hospital is there because of a genetic problem.
- Each of us carries an average of between four and seven abnormal recessive genes.

Moreover, it is now recognized that the major cripplers and killers of adults – diabetes, heart disease, various psychiatric illnesses, and some cancers – all have significant genetic components. Obviously, then, we all have a tremendous stake in the outcome of current research in the field of human genetics. Fortunately, that research is blazing frontiers almost undreamed of just a generation ago. Hardly a day goes by that one does not read in a newspaper or magazine about some new discovery or breakthrough. In fact, it can be said that nothing less than a revolution is occurring in our understanding of human genetics. The new developments promise to reduce the prevalence of genetic disorders and improve and prolong the lives of their victims. Among the recent advances: more and better tests for prenatal diagnosis; improved technology to sustain the lives of seriously ill and deformed newborns and increase the life expectancy of people afflicted by genetic disorders; and the capacity to undertake mass screenings to identify carriers of harmful genes.

This genetics revolution is already affecting your life, whether you realize it or not. The family doctor who once took your family health history by asking what childhood diseases you had and whether your parents or grandparents had suffered from diabetes or heart disease, has expanded the medical pedigree with questions about your ethnic origins, dates and causes of death of all four grandparents, and the kinds of diseases suffered by other relatives.

An increasing number of knowledgeable people are seeking information about their genetic makeup and risks.

The new human genetics is revolutionizing medical research. It is providing precise information on who is most vulnerable to what kind of illness and who particularly should avoid certain environmental agents. It is helping researchers design more effective and less harmful drugs, and it is providing fresh insights into the function of regulatory genes that affect all human growth and development, from birth to death.

Such major illnesses as heart disease, cancer, and schizophrenia, each of which probably involved many different genes, are too complex to yield their secrets even to the new sophisticated tests and therapies. However, some multi-gene diseases may be prevented or lessened in severity by means of drugs, or by manipulating lifestyle or other environmental variables. As scientists discover the genetic risk factors of certain diseases, people who find out that they are susceptible may be able to reduce their chances of illness simply by altering specific aspects of their behavior and environment. As research advances, it is enabling doctors to help millions of people prevent or control inherited diseases, including some diseases that have been identified only in the last twenty years.

Your family physician has marvelous new tools and data at his disposal for the early diagnosis and treatment of genetic disease, but his success in handling a particular case depends in large measure on the patient's own assistance in acquainting him with the family's health background. Few people, however, are able to give their doctors a comprehensive family health briefing, simply because they have limited knowledge themselves of the family's medical pedigree. If your grandparents died before you were born or while you were small, you may have no idea what diseases caused their deaths, and recollections by other family members may be faulty or not entirely forthright. Most of us are not sufficiently knowledgeable about the medical histories of our other relatives – even of our brothers and sisters. And just knowing the cause of death of family members is not enough, for your grandfather may have died of what was termed “old age” on his death certificate, but he may also have suffered from diabetes or arthritis, which may have been the primary reason for his decline.

Determining a genetic link for a disease can be extremely complex, especially where one inherits the disorder through a recessive gene. Individuals affected with a recessive disease will often find no pattern or precedent for the disease in their family history. While you yourself may not suffer any medical problems stemming from it, if your spouse also carries the same recessive gene, then the disorder can manifest itself in your children or show up in your grandchildren if your children's spouses' also have the same recessive gene.

Many developmental anomalies, birth defects, and disease processes simply seem to “run in families” without being associated with predictable high-recurrence risks. These disorders are referred to as “multifactorial” traits since several factors, both genetic and environmental, are involved in their etiology.

Chromosomal abnormalities comprise another classification of genetic disorders. Some of these may be inherited with a high-recurrence risk, and only through chromosomal analysis of the affected individual and appropriate relatives can persons be identified who may be unaffected carriers capable of passing on the abnormality.

In X-linked (sex-linked) diseases, the characteristic pedigree pattern is that of one or more affected males in multiple generations who are related through apparently normal female carriers – persons who can transmit a genetic disease to their children, but who are not ill with the disease themselves. Hemophilia is an example of this type of X-linked disease. Carrier females are at a 25-percent risk of having affected male children, and all daughters of affected males are obligate carriers. However, some family structures are such that the gene may have been passed through several unaffected females, and, by chance, not inherited by their sons.

## **Your Family Health Tree**

In the diagnosis of any genetic disorder it helps to have as detailed a family history as possible. For example, if you are of Jewish origin, your doctor should know whether you descend from Sephardic, Oriental, or Ashkenazi ancestors, as there are genetic diseases that are characteristically found in one group and not in the others. Frequently it is necessary to examine numerous members of the patient's family before diagnosis of a genetic disorder can be made with certainty. And today, with families scattered throughout the country – even the world – it is unlikely your family doctor would be able to do this; which makes a thorough, documented family health history all the more valuable to you and your relatives. In addition to enhancing the accuracy of diagnosis, a complete family health history also is necessary for effective genetic counseling. Such counseling concerns itself not only with the question of the probability of other family members being affected, but also with aspects of prognosis, treatment, and emotional adjustment. Effective counseling can be achieved only with a full medical history of the family.

A good family health history should include information about you, your siblings, your children, and your ancestors and their siblings. If you are female, your doctor will probably ask you questions about any abortions, stillbirths, miscarriages, children who died during infancy and members of the sibship who have died. A thorough prenatal history is crucial if you are starting your own family. This should include mention of any use of drugs, consumption of alcohol, and exposure to various infectious diseases. You should always be concerned about the presence of congenital anomalies in your family as well as the clustering of any traits or diseases, and tell your doctor about them.

Knowledge of the ethnic background of your family as well as the place of birth of parents, grandparents, and occasionally great-grandparents, is obviously relevant and important information. The places of their births should include not only the country but also the city or region, and when available the name of the hospital. However, few except experienced genealogists can answer all these questions.

Let me offer an example here to illustrate how beneficial the task of reconstructing a family's history can be. An avid genealogist friend called me one day to see if I knew anything about the migration patterns of Italians or the Spanish to Ireland and Scotland, where most of her family had originated. Her six-week-old grandson had been diagnosed as having thalassemia major, and the doctors wanted complete information about the child's family tree.

Thalassemia is a hereditary form of anemia, occurring most often in people of Mediterranean descent. Causing enlargement of the spleen, iron accumulation in the tissues, respiratory difficulties, and retarded growth and development, it is a serious disease with no known cure. The

only treatment for it requires repeated transfusions, and hence is complicated, painful, and costly. “I need to know if any of my Scotch-Irish families might have had roots in the Mediterranean, especially Italy,” my friend said. She had been unable to find any connection but thought that perhaps there had been some migration from the Mediterranean area to the British Isles. So began a search into a pedigree that had been traced to Ireland and Scotland on the maternal side, with very little known about the paternal line of the child. His genealogist grandmother searched diligently to pinpoint the origins of the family, but despite her efforts she could turn up no known connection to any places in the Mediterranean. All of her and her husband’s families had come from the British Isles as far back as they could determine. Information about the paternal ancestry of the child also revealed only northern European roots.

The child’s doctor continued to believe he suffered from thalassemia, and recommended that all the maternal family members be tested for the disorder. However, on the basis of the good geological data provided by the maternal grandmother on the origins of both families involved, they decided to re-test the child, and thereby discovered that an error in the interpretation of the original test had been made. By being able to rule out thalassemia, the doctors then were able to identify and treat the child’s illness.

Family history encompasses more than genealogy, which is simply the tracing of descent. It includes genealogical charts and lineages, but more broadly, it is the story of one’s ancestors – in effect, the sum of their biographies. Today genealogists themselves are more interested in family histories than in bare birth, marriage and death records. They recognize the importance to their clients of including information about the health of ancestors, realizing that while material gathered today may not be of immediate significance, its long-term value could be incalculable. As the science of genetics becomes even more refined in future years, comprehensive and well-documented family health histories will be a boon to your grandchildren and great-grandchildren and later generations in their quest for good health.

In 1984 the largest hereditary society in the nation, the 200,000-plus-member National Society of the Daughters of the American Revolution (DAR), launched a Family Tree Genetics project in conjunction with Vanderbilt University’s School of Medicine at Nashville, Tennessee. The DAR donated \$50,000 for the computer software for the study and its members submitted thousands of five-generation medical-genealogical charts. Sarah Hughey King, then president-general of the DAR and the leading force behind the project, urged members to participate because “early diagnosis of glaucoma, diabetes, growth hormone deficiency, Huntington’s disease, cancer, circulatory disorder, dyslexia and Alzheimer’s, to name only a few [genetic diseases], makes it possible to treat and, in many instances, prevent problems for future generations.” “We are fortunate,” she observed, “in that we know whence we came. Our research will serve as an inspiration to others.” Under the directorship of Dr. John A. Phillips III of the Division of Genetics at Vanderbilt, the research and tabulation of the DAR members’ medical genealogies has continued, with the results scheduled to be published in the near future. [*Editor’s note*, 2007: this project is still in progress.]

## **Early Detection**

It is believed that genetic factors may be involved in 25 percent of diseases. Frightening as this figure is, many genetic disorders once thought to be incurable can now be controlled or treated

successfully if they are diagnosed *early*. Family health histories can provide descendants with an invaluable tool, alerting them to watch for early warning signs of such illnesses as breast cancer, diabetes, and glaucoma. By compiling a family health tree, you can provide clues to medical problems that have plagued your families for generations, and you can alert your descendants to potential health problems – even before symptoms appear. If there are genetic disorders in your family, they can be diagnosed and treated in light of current scientific knowledge. Given the pace of the genetics revolution, you may find that you are able to minimize or prevent some illnesses that doomed family members in the past.

The recent wave of genetic research has already brought about the following discoveries:

- There is a widespread genetic defect which may explain why up to 10 percent of whites in North America and Europe do not properly metabolize certain drugs. It is estimated that between 35 and 43 percent of the white population of North America and Europe carry at least one copy of this mutant gene. A recent study involving the drug debrisoquine, which is used to control high blood pressure, revealed this metabolic abnormality. It is a recessive gene, but this genetic trait affects the metabolism of more than twenty commonly prescribed drugs, including antidepressants, antiarrhythmics (which smooth out irregular heartbeat) and the cough suppressant dextromethorphan.
- Researchers have recently developed a blood test to identify children at risk of retinoblastoma (fatal eye tumors). This disease is highly curable if treated before it has spread outside the eye. Identification of the relevant gene means a blood or tissue test can be administered to those who have developed the disease as children and want to know their risk of passing the gene on.
- Scientists have developed a simple blood test to diagnose alcoholism that may also offer a way to screen people to see if they have inherited a risk of becoming alcoholic.
- Research continues on human gene therapy in which doctors transplant copies of a normal gene into the cells of a patient whose own body lacks the gene or has it in an abnormal form. Eventually this process may be used to cure patients with cystic fibrosis, various forms of hemophilia, and possibly muscular dystrophy.

Impressive as they are, these developments represent only the leading edge of a genetics revolution that is likely to have exponentially more far-reaching results. Within the next ten years, between 2,000 and 3,000 genetic markers will be identified that could lead to predictive tests for thousands of illnesses, from stroke to heart attack to cancer to Alzheimer's disease.

You can't pick your genes – or change them, yet – but knowing which family health problems are likely to have been passed on to you and your descendants can alert you early enough to prevent many of them or minimize their effects. Knowing that a certain disease runs in your family may mean you will have to make adjustments in your eating habits or take appropriate medications or have more frequent checkups. Or you may find your risk of a certain disease is not as high as you feared. In addition, many other conditions, though not inherited in the way genetic diseases are, do tend to occur more frequently in some families than in others, and by researching your family

health tree you will be alerted to these also. Those with serious genetic illnesses in their background will find full information about them helpful in deciding (perhaps with the aid of genetic counseling) whether or not to have children. Family medical information gathered now and incorporated in your family history can help young and still unborn generations take advantage of future medical discoveries.

You may uncover some frightening things in your family's medical past. However, you are just as likely to discover that you come from hardy, long-lived stock. Regardless of what you find, you can use that information to improve the health care of your loved ones. And what better legacy to leave your descendants than a family health tree with its valuable clues to good health?

## **What are Genetic Diseases?**

Genes are the chemical information we inherit from our parents at the moment we are conceived. They determine our biological "constitution". Genes cause us to be similar to our parents, and they control how we grow and what we look like. They also make us resistant to some diseases and prone to others.

Each of us carries some faulty genes.

There are several mechanisms by which genetic defects may be transmitted from one generation to another:

1. A single gene is altered:
  - a. autosomal dominant inheritance, where the trait is inherited from one parent and from the previous generation;
  - b. recessive inheritance, where both parents are unaffected;
  - c. X-linked (or sex linked) inheritance, in which the gene for the characteristic is known to be on the X chromosome;
2. Multifactorial inheritance patterns. Several genes on several chromosomes interact with each other. Unknown environmental influences probably contribute to the expression or physical characteristics of this gene.
3. Chromosome rearrangements, deletions, duplicates. We each have 46 chromosomes in 23 pairs, numbered 1 through 22. The 23<sup>rd</sup> pair differs in males and females. Females have two X-chromosomes, and males have one X-chromosome and one Y chromosome.

Von Hippel-Lindau syndrome is caused by an abnormality in the VHL gene, on the short arm of chromosome three. It is an autosomal dominant gene with widely variable expression. This means that if you don't have the disease, you can't pass it to your children. But if you do have the gene you may have mild, or more significant problems. The disease may be so mild in someone that it may seem to skip a generation. For this reason, medical screening is very important.

## Autosomal Dominant Inheritance

Autosomal means that the gene pair is present in a chromosome pair other than the sex chromosomes, so that both male and female children can be affected. In dominant inheritance, an affected child usually has a parent with the same disorder. When the parent has a dominant gene for a disease, there is a 50-percent risk that each child will receive the gene and manifest the defect, though it may not be evident at birth. There is an equal likelihood that a child will not receive the abnormal gene, and thus that child and his or her children should be free of the defect. There are about 2,000 confirmed or suspected autosomal dominant disorders.

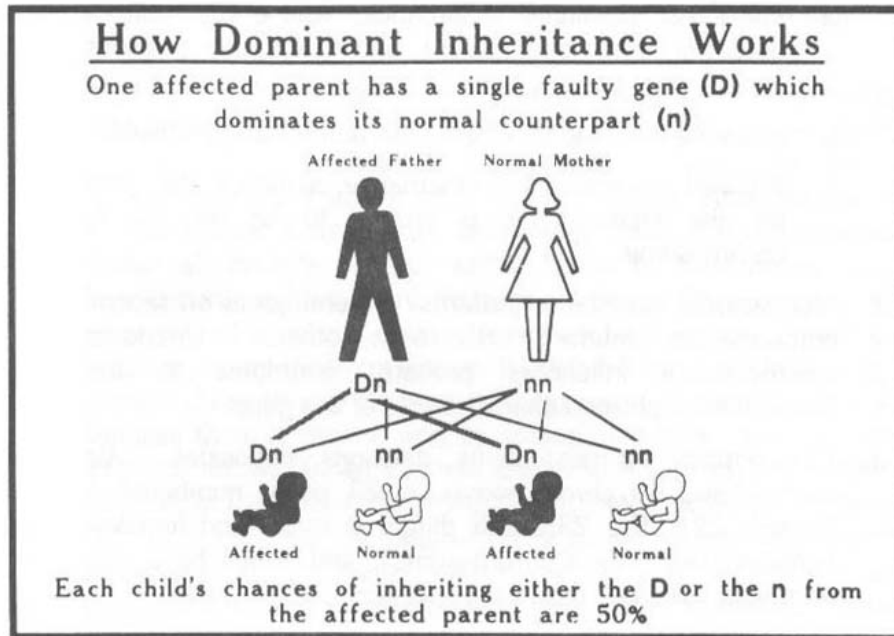


Figure 1: (Courtesy of National March of Dimes)

## Constructing a Medical History Chart

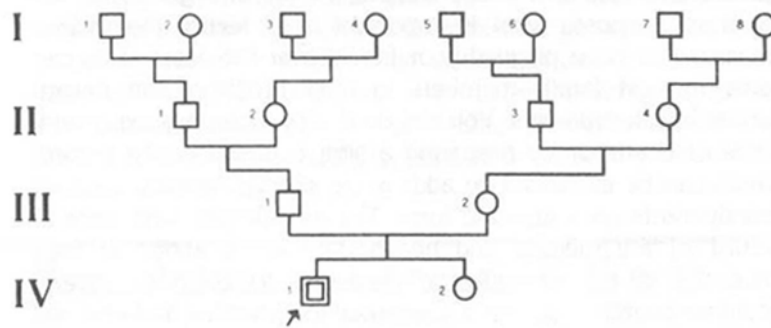
Medical pedigrees like the one in Figure 2, which shows four generations, are normally used to show a particular type of disease or disorder and how it occurs in a particular family. To get an overview of what your family's medical history looks like construct a chart, beginning with your great-grandparents, if known, to show your family members' ages at death and their major health problems and cause of death.

This chart deals with your direct line, and it is an ideal chart to be included in every genealogy. It is often difficult to determine genetic disorders in a family without the aid of a geneticist, but for most purposes what is important is to record the various diseases that have plagued your family over the years. This can serve to alert family members to their predisposition toward certain health problems. You can do this by incorporating several kinds of charts or by preparing a simple family health record, which can be expanded by adding the siblings of each couple's grandparents on a separate form. For genealogists who want to record all the medical and health data found about all their ancestors, it will probably be necessary to construct several pedigree charts or to use a computer to assemble and compile information.

## Asking the Right Questions

It is common to concentrate on the major health problems, like VHL, and to make many erroneous assumptions. It is important to be complete in your information-gathering so that your doctors can see the entire picture.

It will help to ask the right questions. Some questions that are often overlooked of the mothers are: “Are these all of the pregnancies you have had?” and “Are these children all from your present marriage?” Categories that are often neglected include: mental illness, suicide, infertility, extreme tall or short stature, learning disabilities, speech problems and allergies. Other information that is often missed involves medical problems that were treated long ago and forgotten. Possibly family members had repaired hernias or surgery on cleft lip and palate, or congenital heart defects; or there are unrecorded miscarriages and stillbirths. There may have been dental problems. And be sure to ask about any reconstructive surgery that family members may have had.



**Figure 2: Basic Medical Pedigree Chart**

This example covers four generations and shows the basic principles of drawing up a family tree. The eight great-grandparents are shown in Generation I; the four grandparents in Generation II; the two parents in Generation III, and a male proband (also called the Index Person) is shown in Generation IV. Note: siblings in each generation may be added. They would be indicated by an Arabic number from left to right in order of birth.

As a matter of fact, you will probably need to go back to family members a number of times to ask more specific questions. Often the original questions will be too broad and may require more details. It may be useful to have a prepared list of questions. By giving a copy of this list to family members you may help to jog their memories about the family’s health problems. Then you can ask such questions as, “Did Uncle John have any sight or hearing problems? Any unusual birthmarks or moles? Did he suffer from heart disease? Is there anything else that you can think of that gave him health problems? Did he ever have any surgery? What type? Why? Any emotional problems?”

Remember that unless all of your family members are medical doctors or geneticists, the information they provide may be faulty (and even experts are subject to errors) and the terminology may be incorrect. Write down the information exactly as given. However, verify it later with additional research. Aunt Julia may consider herself an expert on the causes of death of family members or about the illnesses of her loved ones, when in fact she may be woefully

incorrect or could have mixed up her information. She may tell you, for example, that grandmother suffered from arthritis, when in fact it was grandpa.

Using this line of questioning you may discover information that was overlooked in the original interview with family members. So after you have constructed your first draft of the pedigree, obtained as much specific information as possible and recorded the documented data (from death certificates, hospital records, etc.), you should go back and ask general questions that cover the entire family.

In looking for possible Von Hippel-Lindau in prior generations, before it would have been identified medically, ask questions like:

- any vision problems? (possible indication of retinal involvement)
- any learning disabilities? chronic headaches? brain tumors or aneurisms? (possible indication of brain involvement)
- any problems with chronic pain? limitations in movement or walking? (possible indication of spinal involvement)
- any cancers? especially abdominal cancers? (possible indication of involvement of kidney, pancreas, adrenals)
- any fertility issues? Cysts or tumors of the reproductive tract in men or women?

Even today, Von Hippel-Lindau is frequently not identified by name, so don't depend on having a clear medical diagnosis from other family members. On the other hand, the presence of these symptoms by themselves does not necessarily mean that the person has or had VHL. Once you have gathered your data, you should begin working with your medical professionals to make the final determinations.

Other Genetic and familial diseases should also be included in your family health tree:

- Alcoholism, allergies, arthritis, asthma, blood diseases (hemophilia, sickle cell disease, and thalassemia); cancer (several forms have shown a familial relationship) – breast, bowel, colon, ovarian, skin and stomach. Some evidence also suggests a familial connection in leukemia and lung cancer;
- Cardiovascular disease (high blood pressure, atherosclerosis, heart attack, hyperlipidemia, stroke, congenital heart defects); congenital abnormalities at birth, cystic fibrosis, diabetes, Down syndrome, dwarfism, epilepsy, hearing disorders, Huntington disease, hypertension, liver diseases (particularly hepatitis);
- Mental illness (particularly manic-depressive disorders, schizophrenia), mental retardation (Down syndrome, PKU); migraine headaches, miscarriages, multiple sclerosis, muscular dystrophy, myasthenia gravis, obesity, phenylketonuria (PKU);

- Respiratory diseases (particularly emphysema, bacterial pneumonia, tuberculosis); Rh disease, sickle cell disease or trait; skin disorders (particularly psoriasis), sudden infant death (SIDS), suicide, systemic lupus erythematosus, thyroid disorders, Tay-Sachs disease; and visual disorders (cataracts, dyslexia, glaucoma, retinitis pigmentosa).

With this information your medical team will be able to get a much to get a much better picture of your family's total health profile.

## **Your Medical Pedigree Chart**

First fill out the Family Health Record included in this booklet as Figure 3. Attach additional sheets as needed to describe all the family members, with notations about diagnosed or suspected VHL along with the other relevant information we have discussed.

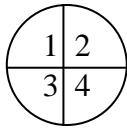
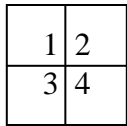
Note the individual and family name at each marriage. If at all possible, diagram the pregnancies from the first to the last in the order in which they occurred, being careful to include all miscarriages and stillbirths. For each numbered individual, fill out the health record as completely as possible, including age at first diagnosis. List all causes of death, if known, and age at time of death.

Please note your relative's country of origin. Family pictures may also be helpful in reviewing your family history.

Figure 3:

FAMILY HEALTH RECORD					
NAME	BIRTH DATE	BLOOD TYPE & Rh	OCCUPATION	DISEASES & INFIRMITIES	IF DECEASED AGE & CAUSE
HUSBAND					
HIS FATHER					
HIS MOTHER					
HIS BROTHER OR SISTER					
HIS BROTHER OR SISTER					
HIS BROTHER OR SISTER					
HIS BROTHER OR SISTER					
HIS BROTHER OR SISTER					
HIS PATERNAL GRANDFATHER					
HIS PATERNAL GRANDMOTHER					
HIS MATERNAL GRANDFATHER					
HIS MATERNAL GRANDMOTHER					
WIFE					
HER FATHER					
HER MOTHER					
HER BROTHER OR SISTER					
HER BROTHER OR SISTER					
HER BROTHER OR SISTER					
HER BROTHER OR SISTER					
HER BROTHER OR SISTER					
HER BROTHER OR SISTER					
HER PATERNAL GRANDFATHER					
HER PATERNAL GRANDMOTHER					
HER MATERNAL GRANDFATHER					
HER MATERNAL GRANDMOTHER					

Once you have gathered your information, map your family on the sample family health tree on a copy of Figure 6, or on your own paper. Color in the circles or squares of people affected with VHL. To further assist doctors and researchers, it is helpful to use the VHL codes suggested here if you have this information.



**Figure 4: VHL Involvement codes:**

black	Clinically diagnosed
striped	Suspected
1	Brain and/or spine
2	Retina
3	Kidney and/or pancreas
4	Adrenal glands

If you are consulting with a genetics professional, it is helpful to send your completed family health records and family health tree in advance of your appointment

Volunteers at the VHL Family Alliance will provide some consulting assistance by telephone, or mail..

## DNA Diagnosis

The VHL gene was mapped in 1993. Since then, DNA testing has moved from the experimental laboratories into the mainstream.

Anyone with a first- or second-degree relative with VHL is "at risk" for VHL. First degree relatives are parents, children, sisters, and brothers. Second-degree relatives are cousins, aunts, uncles, grandparents, and grandchildren. The only way to determine for sure whether someone has VHL is through DNA testing. This is a blood test that must be processed at a clinical testing laboratory (lab) that has the necessary equipment and reagents to test for VHL.

If DNA testing finds the altered VHL gene, we say that the results are positive: yes, this person has VHL. If the DNA testing finds that both copies of the VHL gene are unaltered, we say that the test is negative. This person is unlikely to have VHL. There is always some margin for error. When the possibility of error is under 1-2%, it is considered to be as certain as it gets in nature. If the margin for error is 15%, you may wish to have additional testing.

Anyone at risk for VHL who has not received a negative DNA test result should continue to follow a conscientious screening program to ensure early diagnosis of any VHL problems.

To initiate DNA testing in a family, a person in the family with a clinical diagnosis of VHL, working through a geneticist or genetic counselor, should submit a blood sample for testing. The lab will check to see that they can determine the alteration in this person by performing a complete

screen of the VHL gene. This test is greater than 99% successful in finding mutations in patients with a germline mutation in the VHL gene. Once a mutation has been found, the exact change in this person's VHL gene will be the same alteration that is passed within this family. Now another person in the same family who does not have a clinical diagnosis of VHL can submit a blood sample, and the lab can go directly to that position and check for that same mutation in this second person's DNA. This first test in the family becomes a road map for the second test.

People who were tested prior to 2000 using a method called "linkage analysis" may wish to be re-tested using DNA sequencing or Southern blot analysis. These improved techniques are significantly more reliable. There have been situations where the results of linkage analysis have proven not to be correct.

For people who are the first in their families to be diagnosed with VHL, or for adoptees or others who do not have known blood relatives to assist in the testing, it can take 4 to 6 weeks or more to get results from a complete screen. For people in this situation, it is important to choose a lab with a high "hit rate" or level of success in finding mutations.

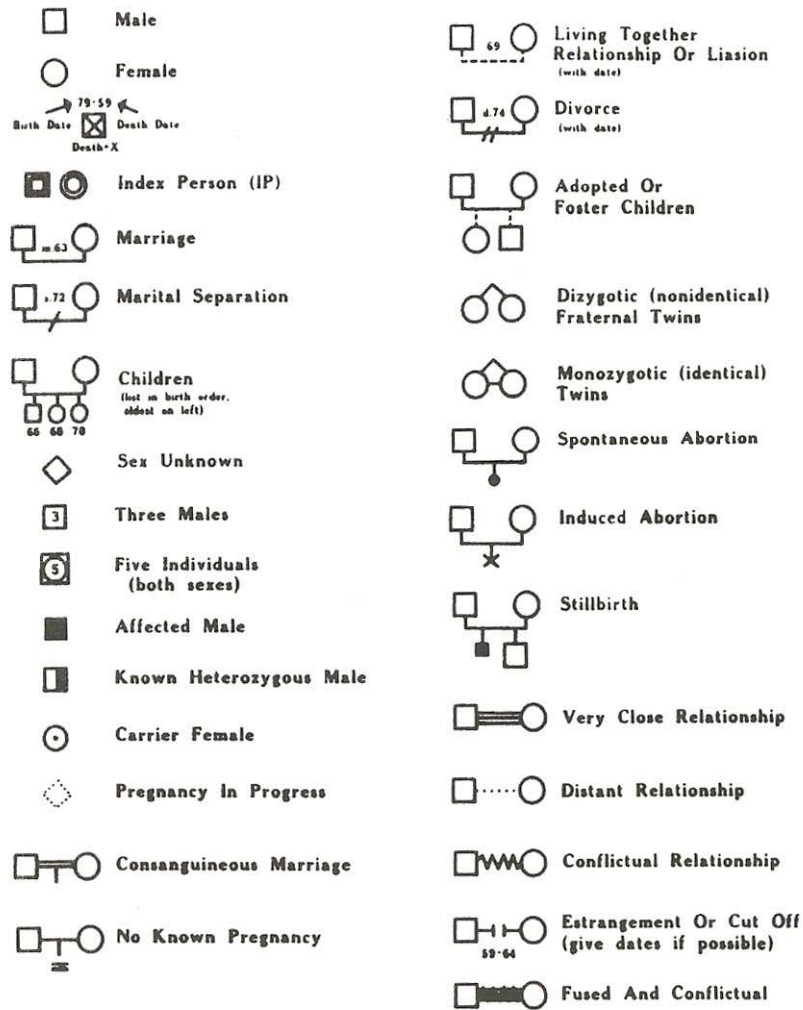
It is important to initiate DNA testing through a geneticist or genetic counselor, to ensure a thorough discussion of the personal impact of the results, whether they are positive or negative, and the possible insurance ramifications. To find a geneticist or genetic counselor, begin with your doctor or with the medical center where you normally go. Ask if they have a department of "cancer genetics." If so, this is the best place to assess your risk for VHL. If not, inquire in the departments of obstetrics, medicine or pediatrics. If they do not have an associated geneticist, they will know where to find one acceptable to your health plan.

If a mother-to-be is having any genetic testing done, she may request a VHL test be part of that scope of tests, especially if there is any VHL in the family at all, or any history of VHL-related tumors in other family members. Prenatal test results are usually part of the mother's medical record, not the child's. Ask to be sure.

The list of clinical testing labs offering testing for VHL is maintained on the internet at [www.vhl.org/dna](http://www.vhl.org/dna). As of the date of publication of this booklet, the labs with the highest "hit rates" are those in Philadelphia, Pennsylvania; Padua, Italy; Saõ Paolo, Brazil; Ingelheim, Germany; and Lyon, France.

Thank You!

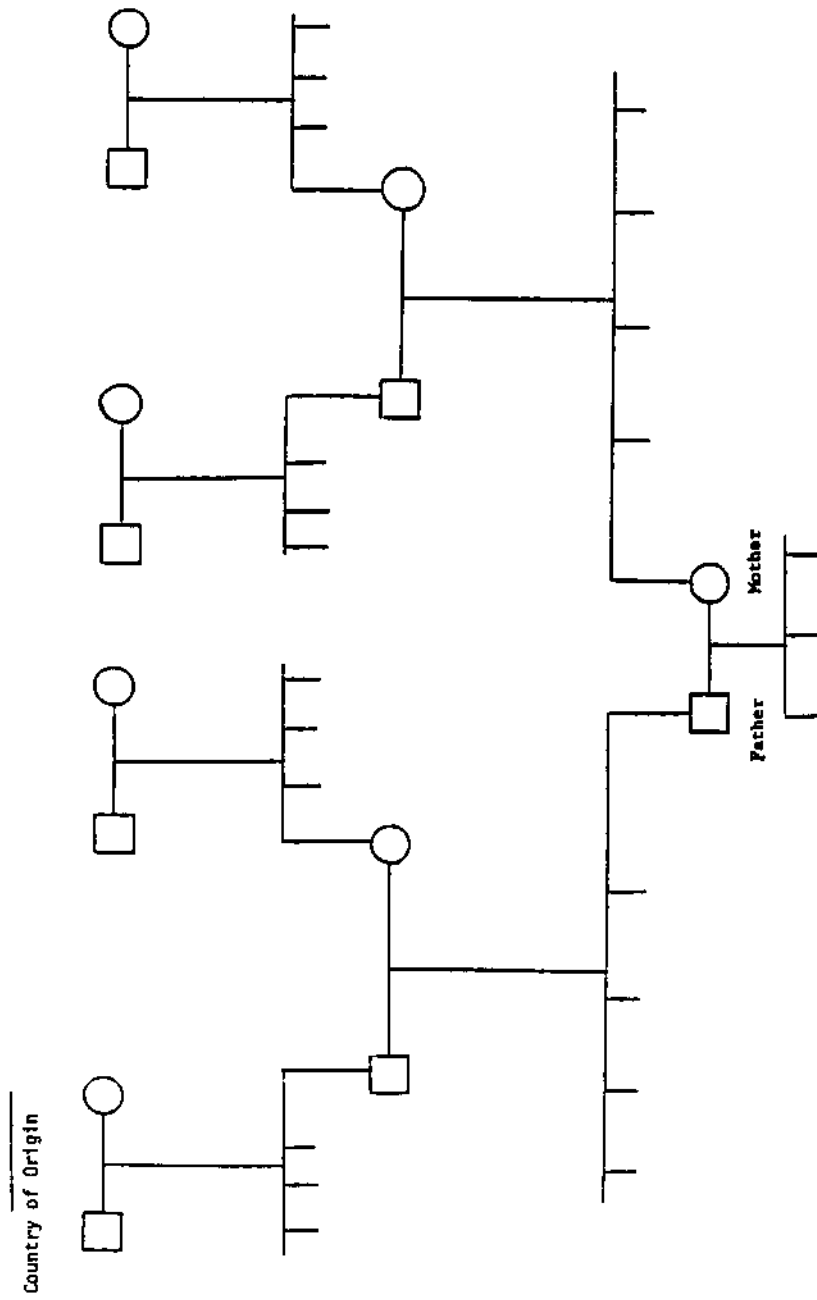
## Genogram Format And Common Pedigree Symbols



*(Courtesy of Kingdom Productions, Houston, Texas.)*

Figure 5

Figure 6



## Suggested Reading

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Information and family support is available from the VHL Family Alliance  
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