

## Visit 5: Genetics and Reproduction

**Objective:** At this visit you have the opportunity to review how PKU affects your decisions about reproduction. Decision-making about “high risk” behaviors will also be discussed.

In this section:

- High Risk Behaviors
- Phenylketonuria and Pregnancy
- *Essentials of PKU*

At this visit:

- Nutrition assessment- bring your 3 day food record to clinic
- Physical assessment- a brief visit with PKU doctor
- Discussion with genetic counselor about PKU, genetics, and reproduction
- Discussion about high-risk behaviors and PKU
  - Smoking, alcohol, drugs, high blood phe levels
  - Effects on decision making (e.g., while driving)
- Confirm next visit

Who is involved:

- **You**
- Your support team:
  - Your parents
  - PKU clinic physician
  - PKU clinic genetic counselor
  - PKU clinic nutritionist



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## High Risk Behaviors

Whether you have PKU or not, there are issues that every individual faces and needs to make choices about. You are in your teens now, but it is important to know that adults of all ages continue to face these choices, too.

Smoking  
Drinking  
Taking drugs  
Having sex

Do your friends smoke? Does drinking occur at the get-togethers you go to? Does anyone you hang around with smoke pot or take other drugs?

You may have already faced situations in which you needed to decide whether or not you would accept a drink, cigarette, or joint. What did you decide? How do you feel about your decision when you look back on the situation?

It's important to know that there are risks associated with each of these behaviors. As your health-care team, it is our job to provide you with reliable information regarding your health and to be clear about our values. We advise you not to smoke, drink or take drugs. We want you to make informed decisions, and act responsibly. We believe that your health and well-being should be a priority and that true friends will respect and support your choices.

As an adolescent, it is your job to take more steps toward independence, to become competent and responsible for your own needs, feelings, and behaviors. You will gradually emerge as a separate, independent person with your own identity and values. The decisions you make on a daily basis result in real consequences that shape your future.

We trust that you will ask for the support you need as you make your own decisions.



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## Smoking Facts

- Cigarettes carry a high risk of cancer.
- Cigarette smoke smells bad.
- Cigarettes are expensive.
- Smoking will decrease your lung capacity.
- Smoking will interfere with your ability to play sports...and your ability to just go for a walk, or run up a flight of stairs.
- Smoking will make your skin age faster.
- Smoking will eventually give you little wrinkle lines all around your mouth.
- Tobacco will eventually stain your fingers and teeth yellow.
- There are 4.8 million deaths worldwide from smoking each year.
- Every 6.5 seconds, someone in the world dies from a smoking-related disease.
- Each year only 4.7% of smokers succeed in quitting.
- 22.3% of high school students smoke.
- 1 out of 3 smokers begin smoking before the age of 14.
- 63% of high school smokers say they want to quit smoking.
- 400,000 people die each year from their own cigarette smoking
- 8.6 million people in the USA currently suffer from smoking-caused illness
- Smokers lose an average of 13 to 14 years of life because of their smoking.

\*Source: [www.tobaccofreekids.org](http://www.tobaccofreekids.org)



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## Drinking Facts

- Alcohol, in any amount, affects your body in some way.
- Alcohol changes the way your brain processes information.
- Alcohol affects your ability to make decisions.
- Drinking alcohol during pregnancy will damage a developing baby.
- Alcohol interferes with your judgment.
- Alcohol can damage your liver.

## Drinking and Having PKU

- High blood phe levels change the way your brain processes information.
- High blood phe levels interfere with your judgment.
- Alcohol changes your brain processing and interferes with your judgment.

### Therefore...

- The combination of high blood phe levels + alcohol dramatically increases these effects. It does not take much. Even one drink paired with high blood phe levels may significantly impair your thinking.

### Also note

- Beer and wine contain phenylalanine.
- Liqueurs that contain cream (e.g. Bailey's Irish Cream) contain phenylalanine.
- Hard alcohol does not contain phenylalanine.

## Know the facts:

**Immediate consequences:** Alcohol affects your mind, body, and self-control. Drinking initially causes decreased attention, alterations in memory, mood changes, and drowsiness. Further consumption may cause feelings of tiredness and confusion, amnesia ("blacking out"), loss of sensation, difficulty breathing, and even death. Additional effects of alcohol include loss of coordination, slowed thinking and reflexes, distorted vision and memory, and poor judgment. Because alcohol compromises your thinking, it can lead to risky behaviors such as engaging in unprotected sexual activity which may expose you to sexually transmitted diseases and/or unwanted pregnancy.

**Long-term consequences:** Alcohol can damage every organ in your body and increase your risk of many life-threatening diseases.

**Safety:** Even if you are not the one drinking, alcohol can affect your safety when people around you are. Those people drinking may be susceptible to violent behavior, car crashes while driving, and other uncontrollable behavior.

**Know the Laws:** In addition to all of the above, drinking alcohol is punishable by state and federal law. In Washington state if you are under 21 years of age, you will be punished for drinking even if you are under the "drunk" or "buzzed" limit. With a Blood Alcohol Concentration (BAC) of as little as .02% – often achieved even after just one drink – you are subject to penalty. Other offenses include simply being in possession of alcohol (Minor in Possession) or having consumed any alcohol (Consumption of Alcohol).

**For more information on legal penalties involving alcohol in Washington State, visit [liq.wa.gov](http://liq.wa.gov)**

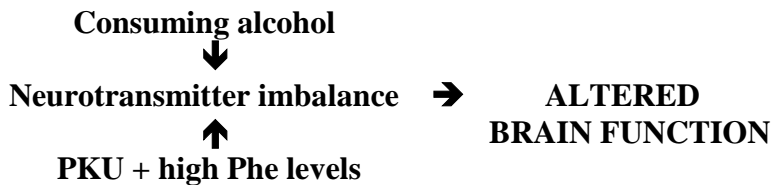


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**Alcohol and PKU:**

Your brain uses chemicals called neurotransmitters to work. They act as messengers that relay information from one cell to the next, resulting in “higher-order executive functioning,” or what we call thinking. In order for your brain to work properly, the balance between different neurotransmitters in different parts of your brain must be maintained. Taking drugs, drinking alcohol, or having high phe levels can all change the levels of neurotransmitters in your brain and keep your brain from working correctly.



**See how having PKU and high phe levels can cause alcohol to effect you differently than others? It is therefore advisable that you be extra cautious about your choices regarding alcohol.**

*\*Photo from <http://blog.longnow.org/2009/07/23/tinkering-with-our-own-brains/>*



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## **Taking Drugs**

- Drugs of any kind will change the way your brain processes information.
- Drugs of any kind, in any amount + high blood phe levels will significantly impair your thinking.
- Drugs that you smoke carry the same risks as smoking cigarettes PLUS the risks of the drugs themselves.
- You can unwillingly take drugs by leaving your drink (non-alcoholic or otherwise) unattended at a party or club. NEVER leave your drink unattended. If you walk away from your drink, get a new one.

## **Marijuana and other illicit drugs:**

- Such drugs alter your mind and consciousness.
- Possessing illicit (illegal) drugs can lead to being expelled or even arrested.
- Many drugs are easily addictive and can lead to life-debilitating consequences.

For more information on drugs, visit [www.drugfree.org](http://www.drugfree.org) and <http://www.abovetheinfluence.com/>



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## Sexual Relationships

Are you in a relationship and thinking about having sex? Are you thinking about having casual sex? This is a topic with several issues, both PKU and non-PKU related issues. Most of the issues are the same for guys and girls. Included in this section is information from Planned Parenthood. The issues about having sex apply both to your first time ever, and also your first time in a new relationship. Sex changes a relationship.

*The Young Woman's Guide to Sexuality*

[www.plannedparenthood.org/womenshealth/ywg.html](http://www.plannedparenthood.org/womenshealth/ywg.html)

*The Young Man's Guide to Sexuality* [www.plannedparenthood.org/health/mangd1.html](http://www.plannedparenthood.org/health/mangd1.html)

*Facts About Birth Control* [www.plannedparenthood.org/bc/bcfacts2.html](http://www.plannedparenthood.org/bc/bcfacts2.html)

## How to practice safe sex:

If you choose to engage in sexual activity, it is important to learn to stay safe and avoid sexually transmitted diseases (STDs) and infections (STIs), and an unwanted pregnancy. This information is intended to be a start to knowing how to have a safe sex life:

- STDs and STIs can be obtained from many different types of sexual activity, including oral, anal, and genital contact with another person as long as one person's genital area is involved.
- The only 100% fool-proof way to avoid any of the unwanted consequences of sex is through abstinence – abstaining from sexual activity.
- Always use a condom. They can be bought at local convenience and/or grocery stores, or often retrieved for free from institutions such as your local Planned Parenthood clinic. Condoms will protect you from both pregnancy and diseases.
- Many women in long-term relationships with a partner that they are regularly sexually active with choose to go on birth-control medications. These include many forms, including a daily pill, periodic shot, or patch worn on the skin. Ask your doctor or make an appointment at a clinic such as Planned Parenthood in order to find out more information.
- Remember that no method (except abstinence) is 100% fool-proof. Condoms can break, and medicated birth control can fail in rare cases. The best way to assure maximum protection is to use these methods exactly according to instructions and consistently (every time you engage in sexual activity).



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- The important thing for a person engaging in sexual activity is that you are educated about your decisions and choosing your own actions. Nobody should force you into doing anything that you would not otherwise choose to do, and if somebody tries to do so it is your responsibility to yourself to take action to put an end to it.

\*This information and more is available at <http://www.plannedparenthood.org/>



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## Sexual Relationships and Issues Specific to People with PKU

There many ways to have a family, whether you are gay or straight. In any kind of relationship where there is to be a pregnancy the information below applies. Having a baby is a huge responsibility for anyone. Even in the ideal scenario, where

1. Both parents are in a committed long-term relationship,
2. The pregnancy is planned and wanted,
3. There is a good household income and health insurance,
4. Neither parent has significant health issues to manage,
5. The baby does not have any special health issues to manage,
6. The new parents have good support systems with friends/family,

...raising a child is HARD, and expensive.

Additionally, if one or both parents have their own health issues that requires planning ahead and organization skills, and/or the child has special needs, the picture gets more complicated and much HARDER.

All couples have a risk of having a child with any significant medical issue at birth. A significant medical issue could be a heart defect, or a kidney problem, or a cleft lip.... The risk for all couples in the general population is about 3-5%. This is your background risk and is separate from your risk of having a child with PKU. Below is a discussion of the genetics of PKU.

## Where does PKU come from?

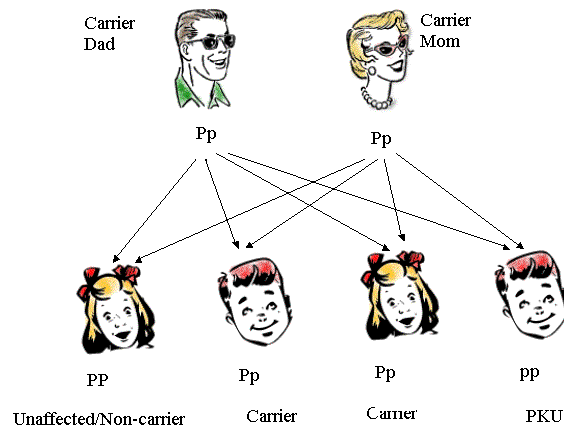
All of our genetic information is stored in the form of genes. Genes are like recipes, which provide instructions to do everything from giving us our hair and eye color, to making the enzymes that break down the food we eat. All of this genetic information is passed down from our parents. We have two copies of every gene: one copy comes from our mother (in the egg) and one copy comes from our father (in the sperm). Individuals with PKU have received two non-functioning copies of the gene for PKU. That is, they have a mutation on each copy. This means that the PKU enzyme (called phenylalanine hydroxylase) made by this gene is not able to function properly and the phenylalanine cannot be broken down. Individuals who receive just one non-functioning copy of the PKU gene are called carriers. Since carriers have one normal copy of the PKU gene, they are able to make enough enzyme to not have symptoms of PKU and never need a low-phenylalanine diet. Couples in which both parents are carriers (like your parents) have a  $\frac{1}{4}$  or 25% chance of having a child with PKU with each pregnancy.



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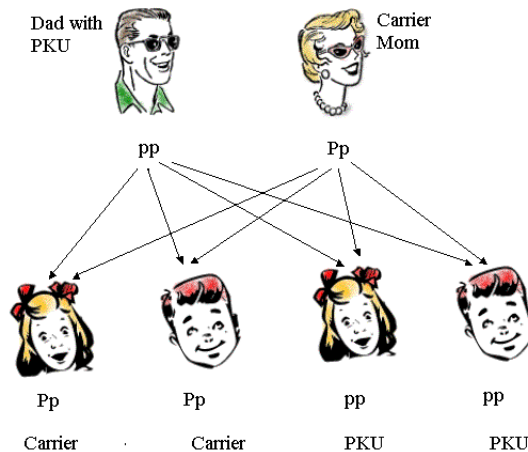
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Legend:  
P = normal gene  
p = PKU gene



### I have PKU. Can my children have it, too?

When individuals with PKU have children, there are two possibilities, depending on whether or not their partner is a carrier for PKU. The person with PKU can only pass on one of his/her mutated genes. If his/her partner is a carrier, he/she has a chance of also passing on his/her mutated gene and the couple will have a child with PKU. There is also a 50% chance of having a child who inherits one mutated gene from the parent with PKU, and the normal gene from the other parent. This child would be a carrier. To simplify things, all of your children will be carriers for PKU. If your partner is a carrier, you have a 50% chance of having a child with PKU.



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## Pregnancy and PKU

High phenylalanine levels in your body will damage your developing baby. The effects vary but generally, the higher the levels, the more damage that happens. Your baby can be harmed by your high phenylalanine levels BEFORE you even know that you are pregnant.

The baby's health problems that result from high blood phenylalanine levels in the mother are called Maternal PKU Syndrome. Maternal PKU Syndrome can include heart defects, a small head (microcephaly), intellectual disability, growth retardation, intestinal defects, and miscarriage (i.e. the damage is severe enough that the pregnancy cannot go to term). A common misconception is that Maternal PKU Syndrome can only happen if the baby also has PKU. The effects of high maternal blood phenylalanine are completely independent of whether or not your child has PKU.

It is very important to use reliable birth control, and use it consistently so that you do not get pregnant before you are ready. Every pregnancy should be a planned pregnancy.



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## Assessment of PKU Adolescent Transition Curriculum

### Session 5.

Objective: At the end of this visit you will have the opportunity to review how PKU affects your decisions about reproduction and socially 'high-risk' behaviors.

Post Session Objective: Each participant will be able to explain how PKU affects his/her decisions about reproduction and socially 'high-risk' behaviors.

I. Please tell us three things that you have learned during this visit

1)

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2)

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3)

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II. How will this information affect your decisions about 'high-risk' activities?

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III. In your view, what is the 'important' decision a person with PKU needs to make about having a family or bearing children?

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