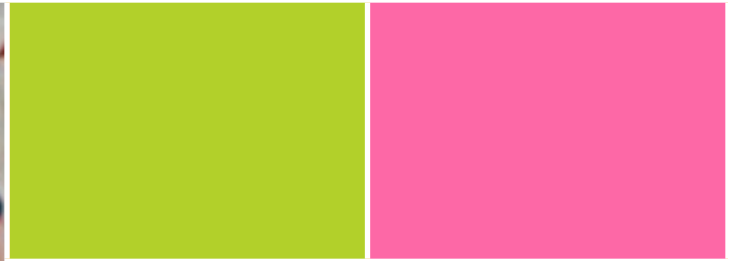


**Moving
Forward**

**Your Guide to
Galactosemia and
Primary Ovarian
Insufficiency (POI)**

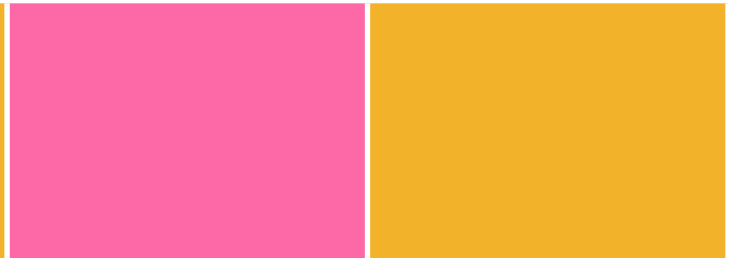


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Most women with galactosemia will face *primary ovarian insufficiency* (POI) at some point in their lives. Some young women with galactosemia and their families find POI to be one of the most worrisome complications of galactosemia. This guide is designed to help you better understand what POI means and learn ways that you can cope with it.

Three key things to remember

1. Neither galactosemia nor POI define who you are.
2. Getting support from family, close friends, and other people with galactosemia can help you cope with POI
3. If you have POI, it is important to take hormone replacement therapy to prevent future health problems.

What is POI?

What you should know about POI and galactosemia



If you were told you or your daughter have POI, *you are not alone*. Most girls or women with galactosemia experience POI at some point in their lives. Young women with galactosemia and their families sometimes have a hard time coping with POI. You likely have a lot of questions about POI and what it means to you. You will find answers to some of your questions in this guide. If you have other questions or need more information, ask your doctor or contact some of the resources listed in the “Other Resources” section of this guide.

What is POI?

POI (sometimes called premature ovarian failure or POF) is a term used by doctors when a woman is under 40 and her *ovaries* are not working normally. This means that her *ovaries* are not making *estrogen* and other hormones that are important to her health. Galactosemia is one of the few known causes of POI. About 1 in 1,000 women without galactosemia have POI. Most women with POI do not know what caused it. Almost all women with galactosemia have POI or develop it at some point in their lives.

What is POI?

What you should know about POI and galactosemia

What health problems are caused by POI?

Estrogen helps women's bones and heart stay strong and healthy. Since their bodies do not produce estrogen, women with POI may be more likely to get the bone disease osteoporosis or have problems with their hearts. This is why it is important to treat POI right away. POI can also cause *infertility*. This means it is very hard for women with POI to have children. However, some women with galactosemia and POI are able to have children at some point in their lives.

What are some symptoms of POI?

When a teenage girl or young women with galactosemia develops POI, she may feel one or more of these symptoms:

- Not starting her period
- Period stopping all of a sudden
- Breasts not growing
- Hot flashes
- Feeling moody or irritable
- Not being able to sleep
- Vaginal dryness



How is POI different from menopause?

You may hear the term “premature menopause,” but this is not correct. The main difference is that menopause describes a permanent loss of ovarian activity but POI may not always be permanent. Some women with POI may produce estrogen or ovulate on and off.

What is POI?

What you should know about POI and galactosemia

How is POI monitored and when should girls and women with galactosemia be tested?

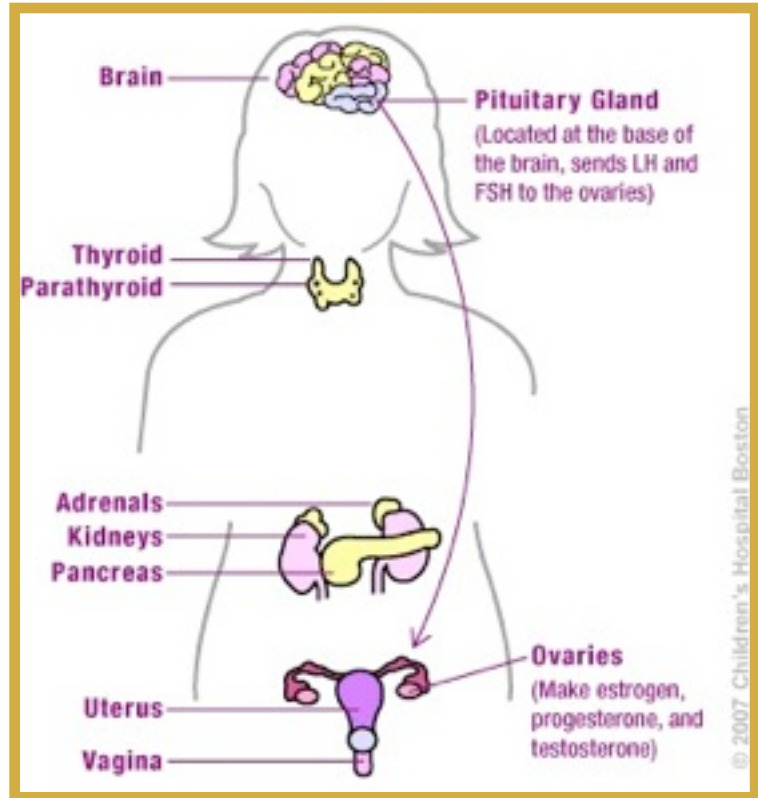
Your doctor will test your *follicle stimulating hormone (FSH)* levels to test for POI. Your doctor will work with you to determine when to start monitoring FSH levels. Many doctors monitor FSH levels in females with galactosemia at one year of age, at the time of puberty (or if there is a delay in puberty), and if menstrual periods become irregular or stop.

Why is a woman's FSH level high and what does that have to do with her ovaries?

To understand FSH, you first need to know that the ovaries produce an important hormone called estrogen. FSH is released by the *pituitary gland*.

The pituitary gland is in the brain and can sense if the ovaries are making the right amount of estrogen. When the pituitary gland senses that there is not enough estrogen being made, it will release FSH to the ovaries signaling them to make more estrogen.

When a woman's ovaries are working normally, they will make estrogen. When estrogen is released, the pituitary gland will stop releasing so much FSH. When a woman has POI, her ovaries cannot produce estrogen. This means her pituitary gland keeps making FSH and her levels of FSH stay very high.



What is POI?

What you should know about POI and galactosemia

How is POI treated?

A girl or woman with POI is treated with medicine that replaces the estrogen and other *hormones* that her body is not making. This type of treatment is called *hormone replacement therapy*, or HRT. Your doctor may start you or your daughter on HRT if there are signs of POI or if puberty is delayed.

When a young woman with POI begins HRT, her breasts and hips may grow if they were not already fully grown. Her period may start for the first time or may start again. She may also have *pre-menstrual syndrome* (PMS) symptoms, such as mood swings. Simply put, her body will act just as if she was making hormones naturally.

There are many types of HRT, such as:

- Pills that a woman will take every day
- Skin patches that deliver hormones through a woman's skin
- Shots that a woman receives on a regular basis
- Soft rings that a woman inserts in her vagina

Most HRT pills contain lactose as a filler. It is important that a woman with galactosemia consults with her doctor to find an HRT that works for her. If she feels any side effects from HRT, she should speak with her doctor right away! Every woman is different. A woman may need to try a couple forms of HRT to find one that works for her.

POI can also be hard to cope with emotionally. If you or your daughter are having a hard time coping with POI, think about speaking to a social worker, a psychologist, a psychiatrist, or another type of mental health expert. You may also want to think about joining a support group or talking to someone else with galactosemia that has been through this situation. If you are not sure where to turn, look at the "Other Resources" section of this guide.

What is POI?

What you should know about POI and galactosemia



Is HRT safe?

You may have heard that hormone therapy causes health problems in middle-aged women who take it after they have menopause. HRT does not cause these problems in young women with POI. The nature of hormone therapy in women with POI is not the same as in women after menopause. Young women with POI are replacing hormones that their bodies should be making on their own during pubertal development or during their early reproductive years. Women who take hormone therapy after menopause are extending the amount of time that their bodies take in estrogen.

One way to think of this is to compare HRT to insulin therapy for people with diabetes. When a woman has diabetes, she takes insulin to replace a hormone her body should be creating naturally. Like the person with diabetes, a woman with POI uses HRT to replace the hormones that her body should be creating on its own.

If you still have questions about the safety of HRT, make sure to talk to your doctor. He or she can talk about HRT in terms of your or your daughter's unique situation.

Can a woman with POI ever have a child?

Most women with POI find it very hard to become pregnant. However, there are some women with POI (around 5% to 10%) who have been able to have children. There is no way to tell which women with POI will be able to get pregnant.

There are many types of families. There are some choices for women with galactosemia and POI who want to have a child. Some women with POI and

What is POI?

What you should know about POI and galactosemia

galactosemia may choose to adopt a child. Other women may want to try *in-vitro fertilization* (IVF), *gamete intrafallopian transfer* (GIFT) or other alternative reproductive therapies. Some women with galactosemia and POI may decide that they will wait to try to have children or that they do not want to have children. Children can still play a key role in a woman's life, even if she does not have children of her own. Every woman with POI and galactosemia is unique and how she (and in some cases her partner) chooses to deal with infertility will depend on her personal situation.

Most doctors say that women with POI should deal with the emotional and other health problems caused by POI before they think about having children. This is one important reason why the parents of an adolescent with galactosemia should not make decisions about their daughters' body or reproduction. Instead, they should wait until their daughter is old enough to make her own decision. If you are ready to learn more, talk to your doctor about the options that you have. Or visit some of the resources listed in the "Other Resources" section of this guide.

Just for parents

Your guide to talking to your daughter about POI



If you just found out about POI in females with galactosemia, you may feel overwhelmed or like this is “just one more thing” about galactosemia that you and your family need to deal with. You may also feel sad that your daughter may not be able to have children. Some parents find it hard to talk to their daughter about POI. Many studies have shown that family support is key to helping young women cope with POI.

You can play a key role in helping your daughter prepare for and cope with POI

For parents

Your guide to talking to your daughter about POI

Five things to think about when it comes to talking to your daughter about POI:

- 1.** Many families of girls with galactosemia learn about the potential of POI when their daughter is very young. This means that you have time to help your daughter prepare for and cope with POI from a young age.
- 2.** Some parents are tempted to keep POI a secret from their daughter. This is most often because they want to protect their daughter from feeling sad or stressed. However, studies have shown that keeping this information from a young woman can, in fact, hurt her ability to cope with POI.
- 3.** It is normal to feel sad, upset, or angry when you learn about POI and galactosemia. However, it is important that you address these issues in yourself early so that your daughter does not sense that you are disappointed or upset. A parent's long term grief about his or her daughter's POI can make it very hard for a young woman with galactosemia to cope with and adapt to POI.
- 4.** One of the top reasons that parents don't discuss POI with their daughters is that they feel they do not have the resources they need to talk about it and help their daughters cope. This guide will help you get ready to talk about POI. If you need more help understanding POI and how to talk to your daughter about it, talk to your doctor or a clinic social worker or psychologist. Some parents also find it helpful to talk to other parents who have been through a similar situation. The "Other Resources" section of this guide lists groups for parents of children with galactosemia and support groups for people with POI.
- 5.** Since girls and women with galactosemia may be dealing with other health problems, it is hard to say exactly when you should start talking to your daughter about POI. You will want to think about your daughter's age, her cognitive skills, and the severity of her galactosemia when it comes to sharing this information. Work with your daughter's doctor and other clinic staff to tell your daughter about POI in an open and honest, yet developmentally appropriate, way.

For parents

Your guide to talking to your daughter about POI

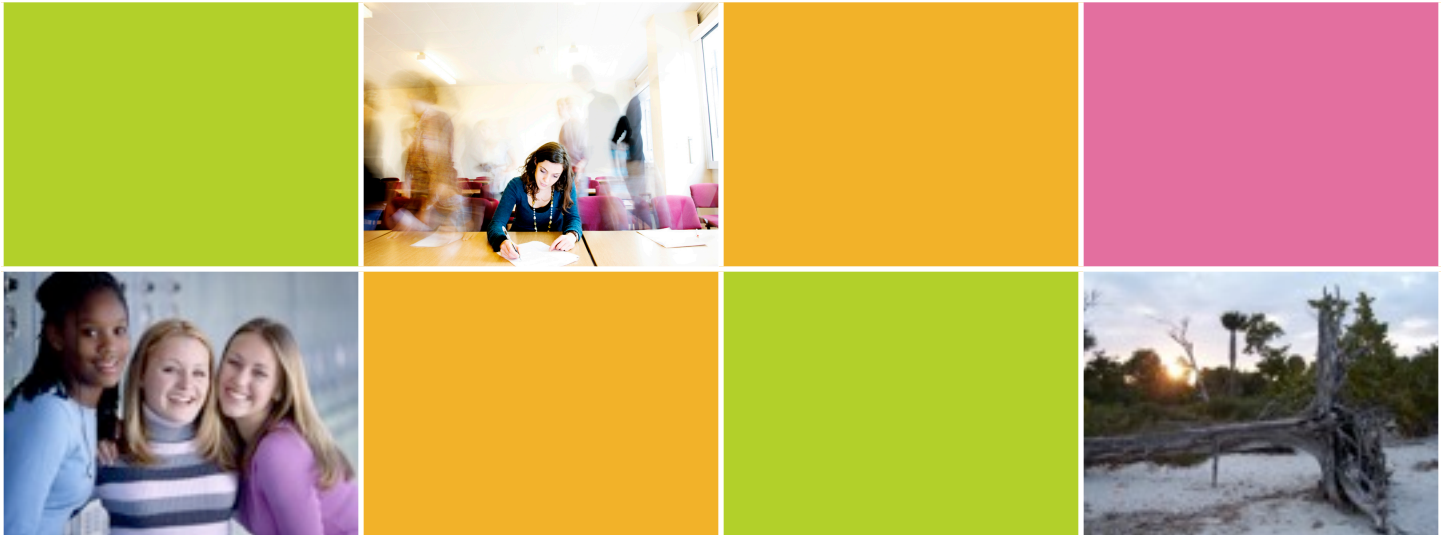
Tips for talking to your daughter about POI

- Look for developmentally appropriate ways to talk to your daughter about POI from a young age.
- Try to create a family setting where a woman's femininity is not defined by having children.
- Show positive examples of alternative ways of becoming a parent, such as adoption.
- Show examples of how children can play a key part in a woman's life even if she does not have children of her own.
- Your daughter may have questions about POI that she is not comfortable asking in front of you. When your daughter learns about POI, you may want to suggest that she spend some time with her doctor by herself. This will also help your daughter get ready to transition to adult healthcare.
- POI is complex. Encourage your daughter to ask her doctor a lot of questions about POI. Tell her that it is okay for her to ask her doctor to repeat something or explain it differently if she doesn't understand.
- Don't leave dad out! POI is a woman's health issue, but leaving her father (or any other key males in her life) out of the discussion may make your daughter feel that POI is something to be ashamed of or something that she shouldn't share with the trusted males in her life.
- In addition to family support, social support helps women cope with POI. Remind your daughter that she is not alone. Encourage her to talk to other women with galactosemia who have also been told about POI.



For women with galactosemia

Your guide to taking action



Learning about POI can be upsetting. You may feel like this is yet one more health problem that you have because of galactosemia. You may feel very sad or upset about this.

The most important thing to remember is that neither galactosemia nor POI defines who you are. There are many other things that are important to you. Don't let POI take over your life.

It is also important to know that there are things you can do to cope with POI. This guide is designed to help you learn about POI and to come up with a plan to cope with your POI.

Getting support

Some women with galactosemia and POI have said that talking to other women who have had POI has been the number one thing when it comes to coping with POI. You may be able to find other women with POI through meetings for people with galactosemia. If you are internet-savvy, you may want to look at discussion groups online. Just remember that anyone can see what you post online! There are also online support groups and telephone support lines for women with POI. Check out the "Other Resources" section of this guide for ideas on where to go for support.

For women with galactosemia

Your guide to taking action

You do not have to go through this alone. Your family members and close friends can also be sources of support. Think about sharing this guide with them so they can learn more about POI.

Getting treatment

It is important that women with POI get treated as soon as possible to prevent health problems. Most women with POI are treated with hormone replacement therapy (HRT). Here are some things to think about when it comes to HRT:



- There are many types of HRT. HRT can come in the form of a pill, a skin patch, a vaginal ring, or a shot. Work with your doctor to decide what type of HRT is right for you.
- If you feel any side effects from HRT or if you do not like the way the HRT makes you feel, speak up! There are many types of HRT and you may need to try a few before you find one that works for you.
- Some HRTs are also used as birth control for women without POI. Even though pregnancies are rare in women with POI, some women do become pregnant. If you are not trying to have a child, make sure that you use a different type of birth control, such as condoms, to prevent unplanned pregnancies.

For women with galactosemia

Your guide to taking action

Talking to your doctor

There is a lot to know about POI, and you will probably have a lot of questions. The way you act can have a big effect on how your doctor talks with you. Here are four tips to help you work with your doctor to learn about POI.

- Some women freeze up or forget what they wanted to ask when they meet with their doctor. It is okay to bring a list of questions with you. You can also ask your parents or another family member to help you remember what you want to ask.
- If your doctor is using big medical words you don't know, ask him or her to be clearer.
- Some people find it easier to learn about POI through visuals. Ask your doctor to use a diagram, a drawing, or a 3-D model of the female body to show you what POI means.
- POI is nothing to be embarrassed about, but you may have questions you'd rather not ask in front of your parents. Ask to meet with your doctor one-on-one. Your parents will understand.



Talking to your partner

It can be very hard to talk to your husband or partner about POI, but it is also very important. Any serious partner should know about your POI so you can come up with a plan for the future together. It is also important that he know so he can be there to support

For women with galactosemia

Your guide to taking action

you. It is hard to say exactly when you should tell a partner about POI, but if you are in a serious relationship you may want to think about these questions:

- When will I tell my partner about POI?
- How open will I be about this?
- What can he do to help me cope with this?

Some women with POI worry that they will not be able to find a loving partner because of their POI. This is not true. Your womanhood and femininity are not defined by your ability to have children on your own. The right person will love you for who you are.

Learning about your choices

It can be hard, but try to deal with the health problems and emotional issues caused by POI *before* you think about having kids. Once you do this and you are ready, talk to your doctor to learn details about the options that you have. He or she may refer you to a doctor who specializes in helping women get pregnant.

Here are some basic facts about some of the choices you have:

- Most women with galactosemia and POI will have a very hard time getting pregnant. However, some women will have children. There is no way of knowing in advance which women will have children. Some couples will decide to wait and see if they get pregnant on their own. If you choose this option, you may want to find out if your partner is a carrier for galactosemia. If he is, your child may have galactosemia.
- Adoption is an option for some young women with POI and galactosemia. There are many children who need a home. Many couples who cannot have a child find adoption to be very rewarding. There are, however, some things to think about. If you are thinking about adoption, it is important to learn as much as you can about the process, the costs, the legal issues, and the possible emotional effects. For more information about adoption resources, look in the “Other Resources” section of this guide.

For women with galactosemia

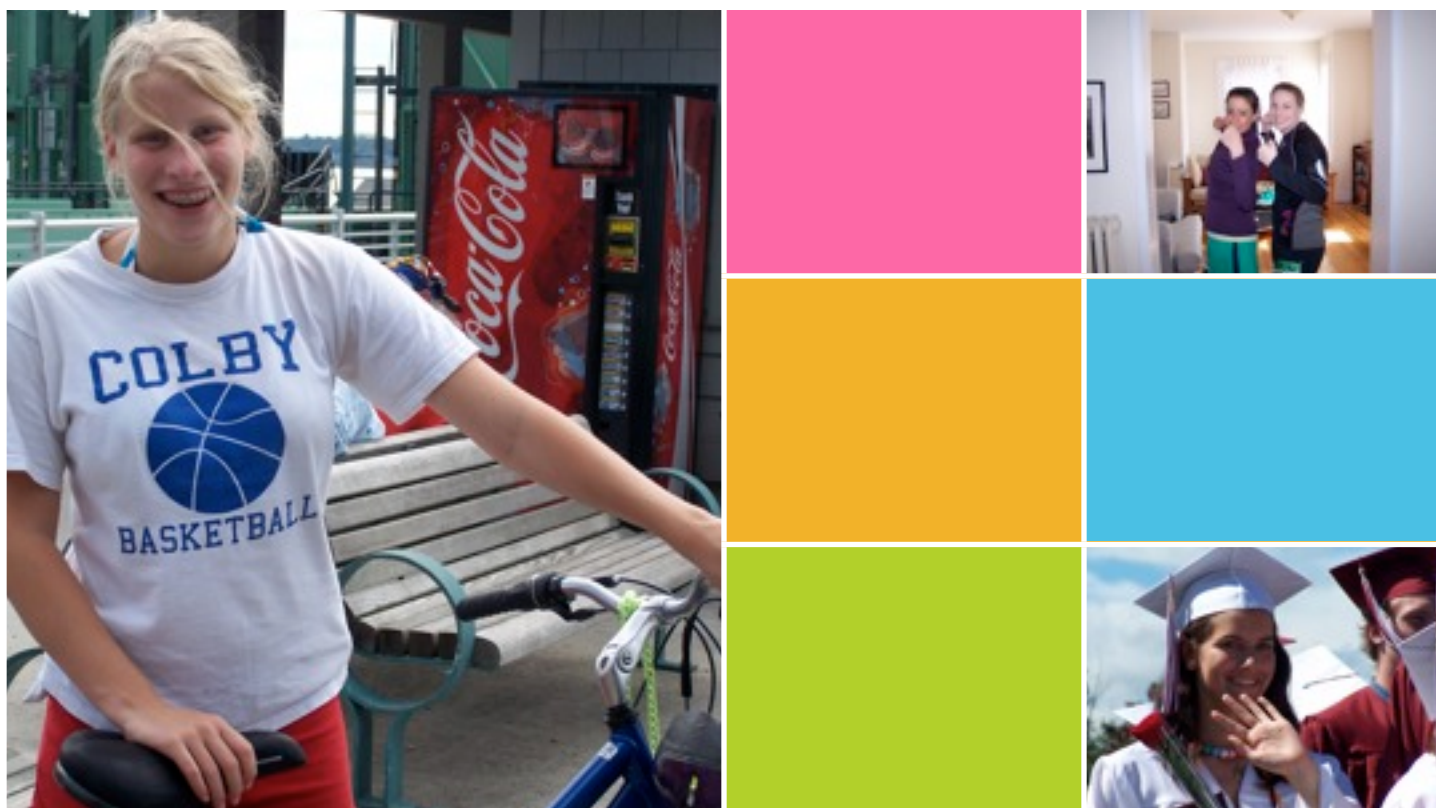
Your guide to taking action

- In-vitro fertilization (IVF) is a medical procedure where an egg from another woman is fertilized with your partner's sperm and then inserted in your uterus. Some women with galactosemia and POI have had success with this method. However, this method can be costly and it may not always work. Make sure you learn about the pros and cons of IVF. For more information about IVF, talk to your doctor or get in touch with one of the resources in the "Other Resources" section of this guide.
- Some women with galactosemia will chose to wait before they try to have children or to live their lives without having children. If this is your choice, remember that HRT does not act as birth control in women with POI. Make sure you use another type of birth control to prevent unplanned pregnancies.

Whatever you decide, it is important that you learn as much as you can about all of your options. The next section includes a list of resources that can help you learn more.

Other resources

Trusted sources for more information



Resources for families affected by galactosemia

Parents of Galactosemic Children (PGC), Inc.

PGC is a non-profit organization dedicated to connecting families affected by galactosemia with each other and with health care professionals. They host a biannual conference that often includes seminars or speakers on POI.

Address:

Parents of Galactosemic Children, Inc.
P.O. Box 2401
Mandeville, LA 70470-2401

Web site: www.galactosemia.org

Other resources

Trusted sources for more information

Galactosemic Discussions

This is a vibrant online discussion forum for people with galactosemia and their families. A woman with galactosemia and her husband created this discussion forum. It covers many topics related to galactosemia and there are several threads that talk about POI.

Web site: <http://www.galactosemics.org/>

New England Consortium of Metabolic Programs at *Children's Hospital Boston*

One of the goals of the Consortium is to develop social support programs and educational materials for patients with metabolic disorders and their families. The "Patients and Families" section of their Web site contains many educational materials and links to other resources.

Phone: (617) 355-7346

Web site: www.childrenshospital.org/newenglandconsortium/Patientsfamilies.html

Resources for women with POI

International POF Association

IPOFA is a nonprofit organization dedicated to providing community, support and information to women with POI (they call it POF) and their loved ones; to increase public awareness and understanding of POI, and to work with health care professionals to better understand this condition. This organization's Web site includes an active discussion forum.

Address:

IPOFA
PO Box 23643
Alexandria, VA 22304

Support Line: (703) 913-4787

Web site: <http://www.pofsupport.org>

Other resources

Trusted sources for more information

RESOLVE: The National Infertility Association

RESOLVE.org is a community for women and men with infertility and provides information, support, and opportunities to take action.

Address

RESOLVE: The National Infertility Association

1760 Old Meadow Rd., Suite 500

McLean, VA 22102

Phone: 703.556.7172

Web site: <http://www.resolve.org/site/PageServer>

RESOLVE of the Bay State Chapter

This is a branch of RESOLVE that specifically serves the entire New England area.

Address:

RESOLVE of the Bay State

395 Totten Pond Road, Suite 403

Waltham, MA 02451

Phone: 781-890-2225

Web site: www.resolveofthebaystate.org

Premature Ovarian Failure: A Guide for Teens

This is an information sheet created by the Center for Young Woman's Health at Children's Hospital Boston. It provides information especially for teenage girls who are dealing with POI.

Web site: <http://www.youngwomenshealth.org/pof.html>

Do I have Premature Ovarian Failure?

This booklet for women and their families explains POI, its possible causes, its symptoms, and its treatments and explains some NICHD research on the condition.

Web site: <https://www.nichd.nih.gov/publications/pubs/pof/index.cfm>

Other resources

Trusted sources for more information

Adoption resources

The Child Welfare Information Gateway

The Child Welfare Information Gateway is a great resource on all aspects of adoption. It is supported by the Administration for Children and Families (ACF), part of the federal government's Department of Health and Human Services. Child Welfare Information Gateway services include technical assistance, a library collection, publications, databases on adoption resources, and information on federal and state legislation related to adoption.

Address:

Child Welfare Information Gateway
Children's Bureau/ACYF, 1250 Maryland Avenue, SW, Eighth Floor, Washington, DC
20024

Phone: 1-800-394-3366 or 703-385-7565

Fax: 703-385-3206

E-mail: info@childwelfare.gov

Web site: <http://www.childwelfare.gov/>

Alternative reproductive therapy resources

The American Society of Reproductive Medicine

The American Society of Reproductive Medicine (ASRM) is an organization devoted to advancing knowledge and expertise in reproductive medicine and biology. ASRM produces many facts sheets and booklets for people facing fertility problems.

Address:

American Society of Reproductive Medicine
1209 Montgomery Highway, Birmingham, AL 35216-2809

Phone: (205) 978-5000

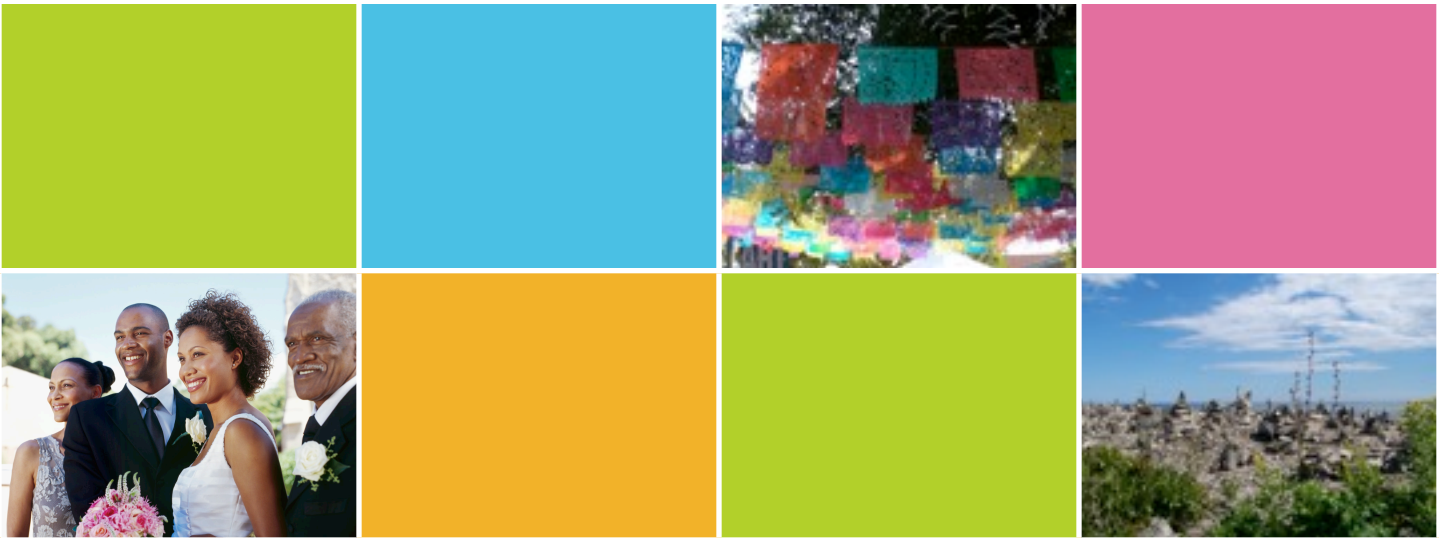
Fax: (205) 978-5005

E-mail: asrm@asrm.org

Web site: <http://www.asrm.org>

Words to know

A POI glossary



Amenorrhea [ey-men-uh-ree-uh]: A word used to describe when a woman's menstrual period is not normal. *Primary amenorrhea* describes when a woman's period does not start when it should (by 15 years of age). *Secondary amenorrhea* describes when a woman's period stops after she has already had it several times.

Estradiol [es-truh-dahy-awl]: An important form of estrogen. It plays a role in the growth of the female uterus and vagina. It also helps breast development.

Estrogen [es-truh-juh-n]: This is a word for several female sex hormones, such as estradiol, that are made by the ovaries. A lack of estrogen can cause health problems like heart disease and osteoporosis.

Follicle Stimulating Hormones (FSH) [fol-i-kuh-l stim-yuh-lat-ing hawr-mohn]: This is a hormone released by the pituitary gland when the brain senses that not enough estrogen is being made. Women with POI tend to have a high FSH level since their ovaries are not able to respond to FSH and produce estrogen.

Gamete Intrafallopian Transfer (GIFT) [gam-eet in-tra-fuh-lop-i-an trans-fur]: A medical procedure where a woman's egg is placed in one of her fallopian tubes with a man's sperm.

Words to know

A POI glossary

Gonads [goh-nads]: This is a word for the reproductive glands found in humans. The female gonad is called the ovary. The ovary is one of a pair that produces eggs and releases hormones like estrogen.

Hormone [hawr-mohn]: A chemical or substance in the body that controls the activities of certain parts of the body.

Hormone Replacement Therapy (HRT) [hawr-mohn ri-pleys-muhnt ther-uh-pee]: Hormone replacement therapy is a medicine that a woman with POI takes to replace the estrogen and other hormones her body is not producing naturally. There are many forms of hormone replacement therapy, including pills, patches, shots, and vaginal rings.

Hypothalamus [hahy-puh-thal-uh-muhs]: An area of the brain that controls things like sleep patterns, hunger, and body temperature by producing hormones, including one that regulates FSH release.

Infertility [in-fur-tl-i-tee]: The diminished ability to become pregnant and have a child. It is also defined as the inability to become pregnant after a year of regular intercourse without birth control.

In-Vitro Fertilization (IVF) [in vee-troh fur-tl-uh-zey-shuhn]: A medical procedure that occurs in a lab where a sperm is placed in a petri dish with a woman's egg. The fertilized egg is then inserted into a woman's uterus.

Osteoporosis [os-tee-oh-puh-roh-sis]: Osteoporosis is a disease in which the bones become weak and are more likely to break. People with osteoporosis most often break bones in the hip, spine, and wrist. Estrogen is important to build strong bones. It is important that women with POI receive HRT to reduce their chances of osteoporosis.

Ovary [oh-vuh-ree]: This is the female gonad. The ovary is one of a pair of reproductive glands in women. They are located in the pelvis. The ovaries produce eggs (ova) and female hormones.

Words to know

A POI glossary

Pre-menstrual Syndrome (PMS) [pre-men-stroo-uhl sin-droh-m]: A combination of physical and mood disturbances that happen before a woman's period. Symptoms can include breast tenderness, bloating, and psychological changes such as anger and depression.

Primary Ovarian Insufficiency [prahy-mer-ee oh-vair-ee-uhn in-suh-fish-uhn-see]: POI (sometimes called premature ovarian failure or POF) is a term used by doctors when a woman is under 40 and her ovaries are not working normally. This means that her ovaries are not making estrogen and other hormones that are important to her health. Galactosemia is one of the few known causes of POI. About 1 in 1,000 women in the United States under the age of 29 have POI. Most women with POI do not know what caused it.

Uterus [yoo-ter-uhs]: A hollow organ found in a woman's lower abdomen. This is where a fertilized egg implants and grows. It is also called a womb.



Notes:

Words to know

A POI glossary
