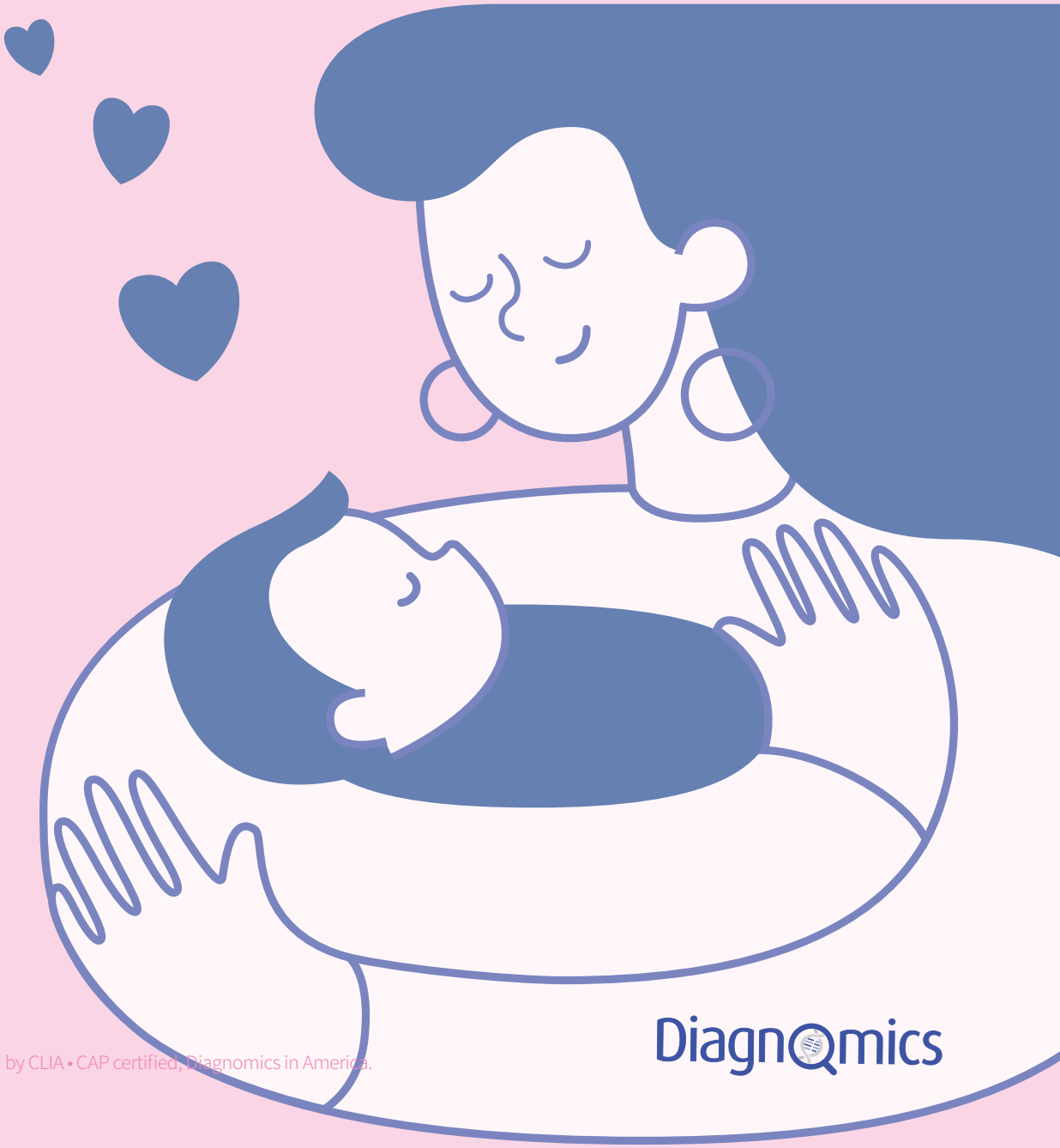
EDGC Momcare

Genetic Test Result Report

• Hospital Name	EDGC
• Examinee ID	A19ddld8fda
• Examinee Name	Hong Gildong
• Client Sample ID	A19ddld8fda



* This analysis was conducted by CLIA • CAP certified, Diagnomics in America.

Diagnomics

Pregnancy Related Items

Diagnomics

[This analysis was conducted by CLIA • CAP certified, Diagnomics in America.]

EDGC Momcare
Genetic Test Result

Contents

About Genetic Testing and Assay QC Results

About Genetic Testing

The genetic information of our body is contained in the DNA. Every person's DNA is mostly identical, but there is a slight difference for each individual. Everyone's genome, however similar, contains slight variations known as SNPs(single nucleotide polymorphisms). These variations are potentially responsible for individual differences, including inherent predispositions to genetic diseases or physical conditions of the individual. This test analyzes individual genetic information with SNP microarray technology and provides information on each items. The result can be used as a reference for understanding the genetic characteristics of each individual.

What Is SNP Microarray?

Single nucleotide polymorphism microarray(SNP microarray) analysis is a technique used to check the single nucleotide polymorphism existing in DNA. This test analyzes the single nucleotide polymorphism through the SNP microarray chip which is integrated with about 700,000 Probes. Because the SNP microarray chip also contains the probe gene for the quality control of the test, quality control and the test itself can be done at the same time. With the SNP test regions used in this test selected from the latest research and accumulated know-hows, we are confident of the accuracy and reliability of this test.

Limitations of the Test (Disclaimer)

This test analyzes the genetic information of an individual and provides information on related items. However, genes not included in this analysis and other environmental factors may also affect conditions, so there can be differences between this test result and the current condition. Examinations and closer inspections by specialists are needed to accurately determine the current status of the examinee. In addition, the result may not be obtainable for less than 1% of the markers used in the SNP microarray analysis, and such genotypes are not shown in this case.

Test Suitability Results

Sample Relevance	DNA Quality	Array Quality	Standard Substance Analysis Result
Compatible	Compatible	Compatible	Compatible

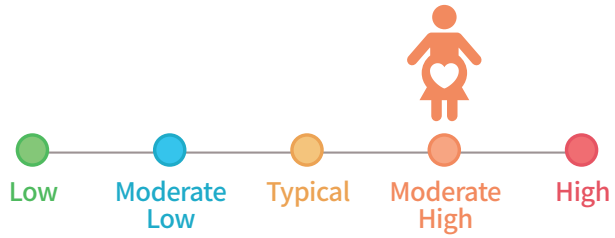
Diagnomics

* This analysis was conducted by CLIA • CAP certified, Diagnomics in America.
5795 Kearny Villa Rd. San Diego, CA 92123 USA | info@diagnomics.com
CLIA ID: 05D2103644 | CAP ID: 9050024 | Lab ID Number: CLF 00348209

Test Results Specification

Genotyping Results and Risk Assessment

• It shows the genetic tendency of examinee on the list.



❶ Genetic Risk: Moderate High
❷ Total Relative Risk: 62%

❶ Genetic risk

The genetic risk is classified into 5 levels of Low, Moderate Low, Typical, Moderate High, and High. The results are predicted based on the research resources and individual genotype results.
- Low Genetic risk is relatively low
- Moderate Low Genetic risk is moderately low
- Typical Genetic risk is relatively lower than slightly high
- Moderate High Genetic risk is moderately high, requiring attention in health care
- High Genetic risk is relatively high, requiring intensive health care

❷ Total relative risk

It shows ❺ **relative risk** of examinee for the item as a percentage(%). The closer the value is to 100%, the more management is needed.

❸ Related Gene (Chromosome Location)	❹ My Genotype (Risk Level)	❺ Relative Risk	❻ Genotype Frequency in East Asian
F11 (4q35.2)	CT Slightly High	1.2	CC Typical 49.39% / CT Slightly High 42.13% / TT High 8.48%

● Risk allele marked in pink

❸ Related gene (Chromosome Location)

It shows the name of gene and chromosomal location, associated with the test site(SNP).

❹ My Genotype(Risk Level)

It is the test result of the genotype analysis of the examinee. The genotype is composed of a pair of genetic elements from parents. There are four genetic factors: Adenine(A), Guanine(G), Thymine(T), and Cytosine(C). The pink letter is a risk factor, related to increasing the risk. The risk is the degree of danger, represented by the examinee's genotype for that item.

❺ Relative Risk

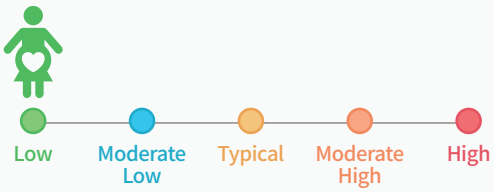
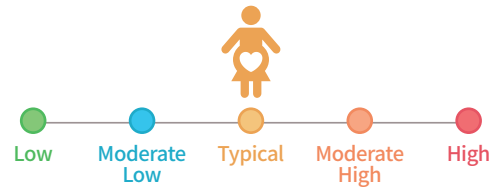
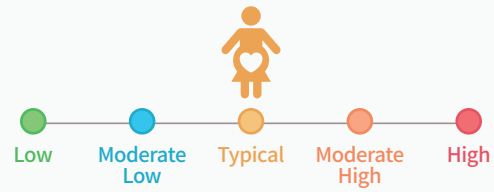
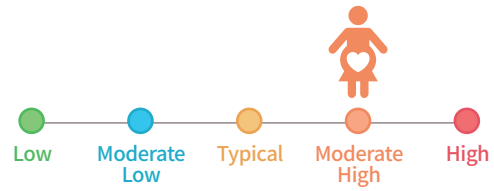
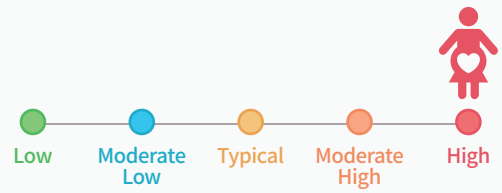
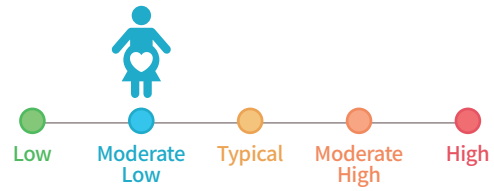
It shows the numerical value of genetic risk, represented by examinee's genotype about the item. The higher the value, the higher the risk.

❻ Genotype Frequency in East Asian

It represents the percentage of each three genotypes in the test site among East Asian population. The corresponding part of the examinee is shown as gray. The three genotypes are listed from left to right in ascending order of risk.

Comprehensive Analysis Result

EDGC Momcare Comprehensive Analysis Result

Item	Total Relative Risk		Analysis Result
Miscarriage - Recurrent Pregnancy Loss: Hereditary Thrombophilia	30%	1.00 (Reference Range 1.31~1.95)	
MTHFR Gene	49%	1.30 (Reference Range 1.01~1.50)	
Obstetric Complications: Thromboembolism	42%	1.20 (Reference Range 1.18~1.50)	
Preeclampsia	62%	1.80 (Reference Range 1.01~1.50)	
Type 2 Diabetes on Newborn's Weight	86%	1.58 (Reference Range 1.11~1.32)	
Preterm Birth	40%	1.00 (Reference Range 1.01~1.50)	

※ The reference range represents the numerical range of corresponding typical level of each item.

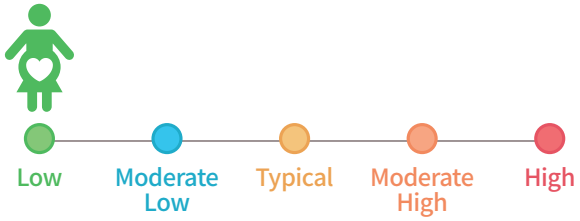
This test predicts the genetic risks. The result may change due to genetic factors, life style, and other environmental factors that are not included in the genetic test result.

Miscarriage - Recurrent Pregnancy Loss: Hereditary Thrombophilia

Overview

Recurrent pregnancy loss refers to cases in which the spontaneous abortion occurs more than three times in a row before 20 weeks of pregnancy. This symptom is caused by several factors such as hypothyroidism, **hereditary thrombophilia**[1], and diabetes. In particular, hereditary thrombophilia, which is closely related to the recurrent pregnancy loss, is a condition in which blood clotting factors become abnormal and blood vessel thrombosis is likely to occur. Thrombosis can interfere with blood flow, inhibiting the fetus from getting enough blood, which can lead to fetal growth retardation and spontaneous abortion. Studies have shown that *F5* and *MTHFR* genes are associated with hereditary thrombophilia. This report provides you with your genetic information related to recurrent pregnancy loss.

Genotyping Results and Risk Assessment



Genetic Risk: Low
Total Relative Risk: 30%

Related Gene (Chromosome Location)	My Genotype (Risk Level)	Relative Risk	Genotype Frequency in East Asian
F5 (1q24.2)	CC Typical	1.0	CC Typical 100.0% / CT Slightly High 0.00% / TT High 0.00%
MTHFR (1) (1p36.22)	CC Typical	1.0	CC Typical 82.35% / CT Slightly High 16.77% / TT High 0.88%
MTHFR (2) (1p36.22)	AA Typical	1.0	AA Typical 67.61% / AC Slightly High 28.93% / CC High 3.46%

● Risk Allele marked in pink

This report only provides results from genetic factors and does not include extrinsic factors, including environmental factors and health conditions. Besides hereditary factors, hormonal factors, immunological factors, infectious factors, and anatomical factors are involved in the occurrence of recurrent pregnancy loss. Parental chromosomal abnormalities account for about 30 to 50% of total recurrent pregnancy loss. Therefore, your final result may differ depending on extrinsic factors.

1) Ford et al. 2009. Recurrent pregnancy loss: etiology, diagnosis, and therapy.

Miscarriage - Recurrent Pregnancy Loss: Hereditary Thrombophilia

Related Genes

F5 Gene

Factor V, produced by the *F5* gene, is involved in the regulation of blood clotting. People with a specific mutation in the gene(Factor V Leiden) may have increased risk of thrombophilia, and the mutation is known to be associated with the recurrent pregnancy loss.

MTHFR Gene

The methylenetetrahydrofolate reductase(*MTHFR*) gene produces enzymes involved in essential amino acid metabolism. If the activity of the enzyme is lowered, the homocysteine concentration is increased. Elevated homocysteine levels in the blood can damage the lining of blood vessels, increasing the risk of thrombosis, which is closely related to recurrent pregnancy loss. Studies have shown that having a specific genotype in the *MTHFR* gene affects the occurrence of recurrent pregnancy loss.

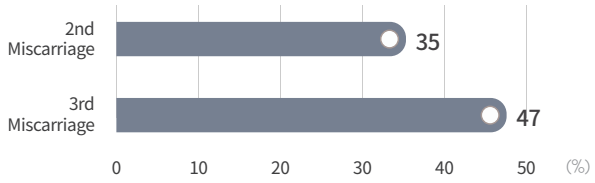
Related Statistics

Incidence of Recurrent Pregnancy Loss

0.5~3.0%

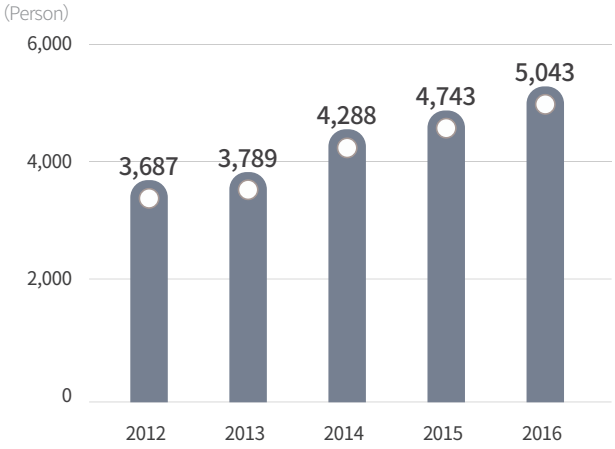
*Source: 2017. Korea Medical Association

Recurrence rate of Miscarriage by Consecutive Miscarriages



*Source: 2016. Korean Oriental Gynecology Journal

Number of Patients with Recurrent Pregnancy Loss by Year

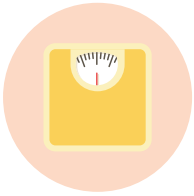


*Source: 2016. Health Insurance Review & Assessment Service

Miscarriage - Recurrent Pregnancy Loss: Hereditary Thrombophilia

How to Prevent Recurrent Pregnancy Loss?

By adjusting lifestyle, you can lower the risk of recurrent pregnancy loss. Recurrent pregnancy loss can be caused by a variety of causes, and the treatment method may vary depending on the cause. Therefore, it is important to consult with a doctor to determine the exact cause and get proper treatment before the next pregnancy.



Weight Control

Overweight or obesity in pregnant women has been shown to increase the risk of recurrent pregnancy loss. It is desirable to maintain proper weight.

Caffeine / Alcohol Ingestion Continence

The risk of miscarriage tends to increase for women who consume excessive caffeine around the time of pregnancy or who regularly drink alcohol during pregnancy. Therefore, it is better to limit the ingestion of caffeine and alcohol.

Emotional Stability

Emotional impacts such as stress or depression can have a negative influence on the occurrence of recurrent pregnancy loss, and constant counseling can help prevent it. Adequate emotional support is essential for couples who have experienced a recurrent pregnancy loss, especially those with unclear causes.

Did You Know?

- 15~25% of pregnant women experience a natural miscarriage.
- Pregnant women over 40 years old are more likely to have a miscarriage.
- The causes of recurrent pregnancy loss are not all determined, and it is not rare that the causes to be classified as unknown.
- Even mothers with experience of recurrent pregnancy loss can have a child with a 60-70% chance. Therefore, it is important to actively make effort for the next pregnancy without giving up.

Miscarriage - Recurrent Pregnancy Loss: Hereditary Thrombophilia

Reference Sites

- **American Society for Reproductive Medicine** https://www.asrm.org
- **Eshre** https://www.eshre.eu
- **Genetic Home Reference** https://ghr.nlm.nih.gov
- **Healthcare Big Data Open System** http://opendata.hira.or.kr
- **Pregnancy Child Care Comprehensive Portal Child Love** http://www.childcare.go.kr
- **The Miscarriage Association** http://www.miscarriageassociation.org.uk

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MTHFR Gene

What Is Folate?

Folate is a type of water-soluble vitamin, vitamin B9, commonly known to be consumed by couples who prepare for pregnancy. However, folate is one of the essential nutrients for both men and women, which plays an important role throughout the entire lifecycle. Deficiency of folate is known to cause various symptoms and disorders by affecting DNA synthesis and cell division. It is especially important for pregnant women because of the serious problems with the fetus including increased risk of premature birth and neural tube defects due to deficiency of folate. Folate deficiency can occur when it is difficult to eat fresh fruit or vegetables. Especially, it is common in people with mutations in the genes involved in folate metabolism, and **about 25-50%**[1)] of the total population is known to have mutated genes.

MTHFR Gene and Homocysteine

The *MTHFR*(5,10-methylenetetrahydrofolate) gene makes MTHFR enzyme which acts on the stage of active folate (5-methylenetetrahydrofolate) production during the process of folate metabolism. With specific variants in this gene, the structure of the MTHFR enzyme will change and the enzyme will not function normally. Active folate is involved in the conversion of homocysteine into methionine in the process of amino acid metabolism. When this conversion process does not work properly due to genetic mutation, excess homocysteine accumulates in the body. Homocysteine is the main cause of vascular damage and can cause various diseases including cardiovascular disease.
30 *MTHFR* gene mutations are known, with C677T and A1298C being the most researched and known.

Enzyme Activation(%)

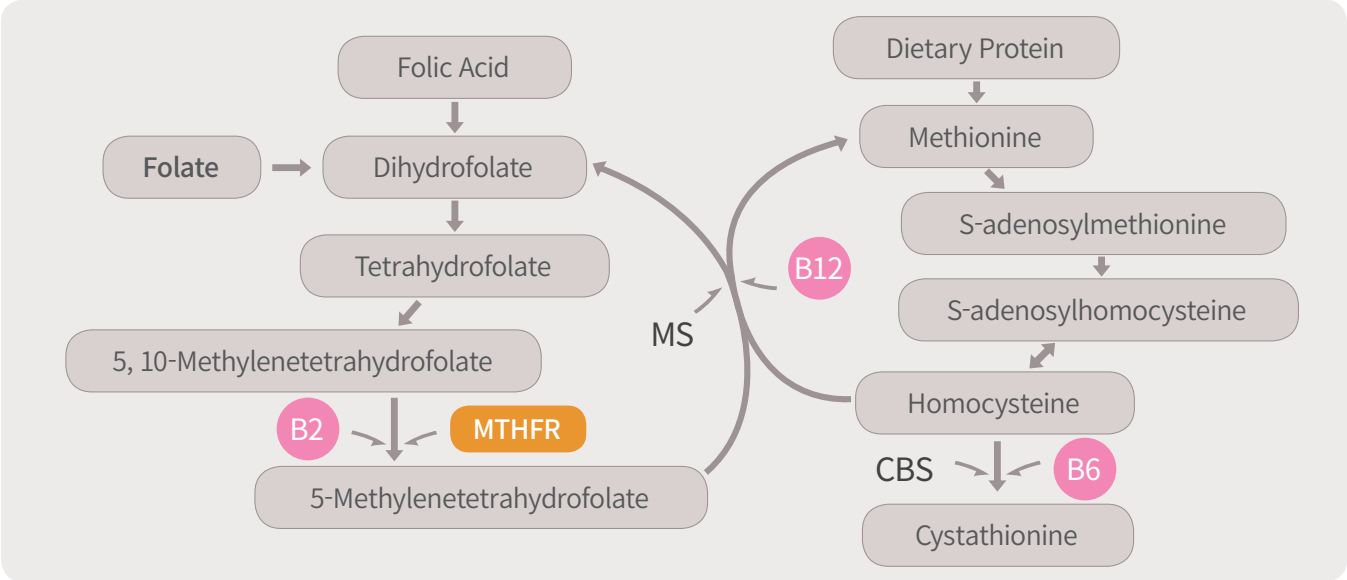
	C677T	A1298C	Compound Hetero
Normal	100	100	.
Hetero	60-70	.	50
Homo	30-40	50-60	.

- Heterozygous: inherited from one parent
- Homozygous: inherited one from each parent
- Compound Heterozygous 677CT + 1298AC: each has hetero combination

1) Wilcken et al. 2003. Geographical and ethnic variation of the 677C>T allele of 5,10 methylenetetrahydrofolate reductase (MTHFR): findings from over 7000 newborns from 16 areas world wide.

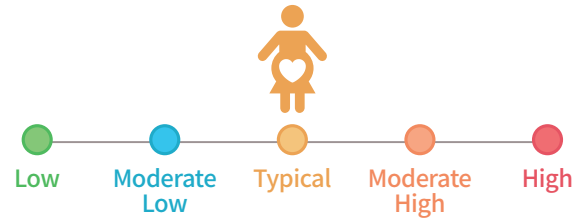
MTHFR Gene

Folate, Homocysteine Metabolism Process



- **MS:** Methionine Synthase
- **CBS:** Cystathionine B-synthase
- **B2:** Vitamin B2 / **B6:** Vitamin B6 / **B12:** Vitamin B12

Genotyping Results and Risk Assessment



Genetic Risk: **Typical**
Total Relative Risk: **49%**

MTHFR Gene Mutation	My Genotype (Risk Level)	Relative Risk	Genotype Frequency in East Asian
C677T (rs1801133)	**CT** Slightly High	1.3	CC Typical 32.17% / **CT Slightly High 50.04%** / TT High 17.79%
A1298C (rs1801131)	**AA** Typical	1.0	**AA Typical 67.61%** / AC Slightly High 28.93% / CC High 3.46%

● Risk Allele marked in pink

This report only provides results from genetic factors and does not include epigenetic factors, including environmental factors and health conditions. Your final result may differ depending on extrinsic factors.

MTHFR Gene

Folate Deficiency? Existence of *MTHFR* Gene Mutation?

Folate is one of the essential nutrients. If deficient, it can be supplemented food, supplements, and injections, etc. In particular, people with mutations in the *MTHFR* gene can benefit from active folate intake.
However, depending on the individual, there is a difference in the amount of folate needed, and side effects may occur when intake excessively. Therefore, it is necessary to consult with a doctor to check the method and amount of supplementation.

* Vitamins B2, B6, and B12 are involved in the metabolism of folate and homocysteine. Therefore, appropriate amount is recommended.

* mcg DFE(micrograms dietary folate equivalents):
Unit used to express the amount of folate.
1 mcg DFE = Folate in food 1 mcg
= Folic acid in dietary supplements consumed with food 0.6 mcg
= Folic acid in dietary supplements with empty stomach 0.5 mcg

Age	Recommended Dose(mcg DFE*)
6 months after birth	65
7-12 months	80
1-3 years	150
4-8 years	200
9-13 years	300
14-18 years	400
19-50 years	400
51-70 years	400
71 years and above	400
Pregnant woman	600
Breast-feeding	500

*USA NIH(National Institutes of Health) Office of Dietary Supplements Recommended Dose

Diseases Associated with *MTHFR* Gene Mutation

More than 60 diseases associated with *MTHFR* gene mutations have been reported. Major diseases are as follows:

Pregnant Woman

- Fetal Down Syndrome
- Fetal Growth Retardation
- Recurrent Pregnancy Loss
- Premature Birth
- Fetal Neural Tube Defect(Spine rupture, aphasia, brain prolapse)
- Fetal underweight

Infant

- ADHD
- Convulsion
- Skeletal Deformation
- Growth Delay
- Decreased Intelligence

Teenager

- ADHD
- Obesity

Adult

- Cardiovascular Disease
- Alzheimer's
- Cancer
- Depression
- Dementia

MTHFR Gene

Folate? Folic Acid?

Folate is a natural vitamin B9 and folic acid is a synthetic vitamin B9 that can be taken as a supplement. Both words find its origin in the Latin folium, which means leaf. As the etymology implies, folate is abundant in green lentils and may need to be supplemented due to individual genetic factors.

Reference Sites

- **Centers for the Disease Control and Prevention** https://www.cdc.gov
- **DIETvsDISEASE** https://www.dietvsdisease.org
- **Everyday Health** http://www.everydayhealth.com
- **Genetics Home Reference** https://ghr.nlm.nih.gov
- **Mayo Clinic** https://www.mayomedicallaboratories.com
- **MTHFR.Net** https://www.mthfr.net
- **Office of Dietary Supplements** https://ods.od.nih.gov
- **WebMD** https://www.webmd.com

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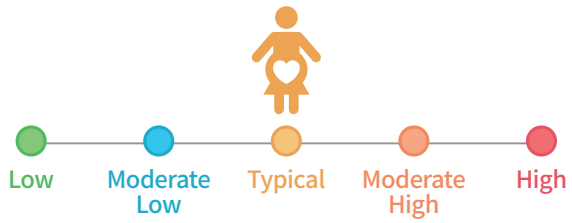
Obstetric Complications: Thromboembolism

Overview

Women experience various changes in their body when they become pregnant. If these changes have a detrimental effect on their health, pregnant women can experience complications such as gestational diabetes, placental abruption, preeclampsia, and thromboembolism. The risk of developing thromboembolism increases by 4 to 5 times during pregnancy, especially during the 3 months of postpartum period. Thromboembolism is a symptom of thrombosis caused by a blood clotting reaction, most of which occurs in the vein. Thromboembolism can easily occur during pregnancy because the blood clotting factor increases and the enlarged uterus compresses the blood vessels, which inhibits blood circulation. In addition to these factors, pregnant women with specific mutations in the *F5* gene and the *F11* gene have shown an increase in the risk of thromboembolism. This report provides your genetic information regarding the *F5* and *F11* genes.

Genotyping Results and Risk Assessment



Genetic Risk: Typical
Total Relative Risk: 42%

Related Gene (Chromosome Location)	My Genotype (Risk Level)	Relative Risk	Genotype Frequency in East Asian
F5 (1) (1q24.2)	CC Typical	1.0	CC Typical 100.0% / CT Slightly High 0.00% / TT High 0.00%
F5 (2) (1q24.2)	TT Typical	1.0	CC Low 4.56% / CT Slightly Low 33.73% / TT Typical 61.71%
F11 (4q35.2)	CT Slightly High	1.2	CC Typical 49.39% / CT Slightly High 42.13% / TT High 8.48%

● Risk Allele marked in pink

This report only provides results from genetic factors and does not include extrinsic factors, including environmental factors and health conditions. Genetic factors explains about 50 to 60% of the incidence of venous thromboembolism, which accounts for most of the thromboembolism, but physical condition and age also involved. Therefore, final result may differ depending on extrinsic factors.

Obstetric Complications: Thromboembolism

Related Genes

F5 Gene

In our body, a blood clotting reaction occurs to prevent bleeding when blood vessels are damaged. The *F5* gene, which creates the fifth blood coagulation factor(Factor V), is involved in regulating this response. People who have certain mutations in the gene(Factor V Leiden) are at increased risk of thromboembolism due to excessive coagulation.

F11 Gene

The *F11* gene is involved in making a protein called Factor XI. This protein circulates in an inactive state and is activated when the blood vessel damaged. The activated Factor XI protein is involved in the production of fibrin, a thrombogenic substance. Certain mutations in the gene are associated with venous thrombosis during pregnancy.

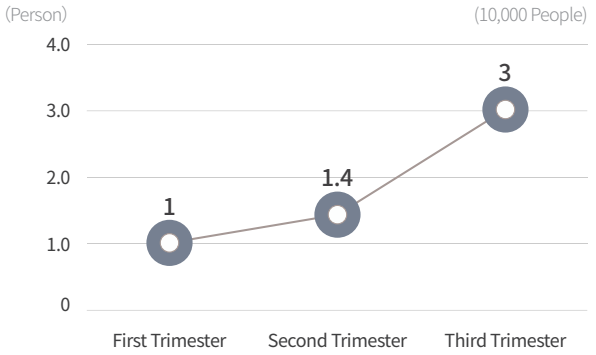
Related Statistics

Incidence of Thromboembolism during Pregnancy

0.7~1.7 cases / 1,000 deliveries

Thromboembolism during pregnancy is a rare disease. According to a study by the Korean Medical Association Journal, the incidence of thromboembolism during pregnancy is 0.7 to 1.7 cases per 1,000 delivery. Of these, 80% are deep vein thrombosis, and pulmonary embolism accounts for 20%.

Number of Patients with Recurrent Pregnancy Loss by Year



*Source: 2016. Korean Medical Association Journal

Obstetric Complications: Thromboembolism

How to Prevent Thromboembolism?

Have Regular Maternity Checkups

Accurate diagnosis is difficult because symptoms such as dyspnea, sensory disturbance, and lower extremity edema are common in thromboembolism as well as in normal pregnancies. Therefore, it is important to regularly consult a gynecologist for comprehensive judgment throughout pregnancy.

Wear Compression Stockings

Wearing medical compression stockings helps blood circulation, alleviating leg edema and numb symptoms, and helps prevent thromboembolism. It is recommended to consult a doctor and get a prescription when getting medical compression stockings.

Sufficient Water Intake

Sufficient water intake prevents blood becoming viscous, which preventing blood circulation problems and thrombosis.

Exercise

Adequate walking and swimming for pregnant women can help improve blood circulation in the legs and help prevent blood clots.

Reference Sites

- **American Heart Association** http://www.heart.org
- **BabyCentre** https://www.babycentre.co.uk
- **Genetic Home Reference** https://ghr.nlm.nih.gov
- **Mayo Clinic** https://www.mayoclinic.org

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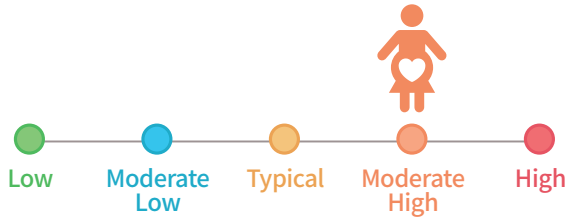
Preeclampsia

Overview

Preeclampsia is a pregnancy complication, related to hypertension. If not properly treated, it can lead to organ dysfunction and other complications. Also, this disease shows several symptoms such as proteinuria, edema, headache, nauseous, vomiting, sharp weight gain, and visual disturbance, etc. Moreover, patients with preeclampsia have a high risk of placental abruption, the separation of the placenta from the uterine lining. The placental abruption is very dangerous to both the mother and her baby. So, in the case of water breaking or abdominal pain, it should be treated at a hospital as soon as possible. According to many studies, although the cause of preeclampsia is unclear until now, genes of the mother can affect this disease at most **35%**[1)] in total.

Genotyping Results and Risk Assessment



Genetic Risk: **Moderate High**
Total Relative Risk: **62%**

Related Gene (Chromosome Location)	My Genotype (Risk Level)	Relative Risk	Genotype Frequency in East Asian
AGT (1q42.2)	**AA** High	1.5	GG Typical 79.19% / GA Slightly High 19.67% / **AA High 1.14%**
AGTR1 (3q24)	**AC** Slightly High	1.2	AA Typical 88.76% / **AC Slightly High 11.11%** / CC High 0.13%
NOS3 (7q36.1)	**GG** Typical	1.0	**GG Typical 98.80%** / GT Slightly High 1.20% / TT High 0.00%

● Risk Allele marked in pink

Preeclampsia is known to affect 3~7% of pregnant women and is one of the causes of premature labor. Risk factors of preeclampsia and early placenta abruption are obesity, diabetes, age, chronic hypertension, multifetal pregnancy (twins, triplets etc.) and a family history. Mutation of *NOS3* gene has been considered as potential risk factors.

1) Saito 2018. Preeclampsia: basic, genomic, and clinical.

Preeclampsia

Related Genes

AGT Gene

AGT(Angiotensinogen) genes are related to the production of protein, called Angiotensin. Angiotensin protein is the part of the Renin-Angiotensin system that controls blood pressure. According to related studies, *AGT* gene is one of the candidate genes related to hypertension and preeclampsia. Specific mutation of this gene can increase systolic/diastolic blood pressure of essential hypertension patients.

AGTR1 Gene

AGTR1(Angiotensin II Type 1 Receptor) gene makes one of the main components in the Renin - Angiotensin system which can control blood pressure. According to related studies, if the *AGTR1* gene has specific mutation, the risk of hypertension onset during pregnancy increases.

NOS3 Gene

NOS3(Nitric Oxide Synthase 3) gene makes an enzyme to help the regulation of blood pressure. The mutation of this gene is related to various hypertension diseases such as preeclampsia, asthma, emphysema and coronary artery spasm. According to several studies, if a woman, who is diagnosed with preeclampsia, has mutations in the specific site of the gene, the potential risk of early placental abruption increases. Also, 49% of women who had experienced with early placental abruption and 21% of women who do not have that diseases have mutations on *NOS3* gene. Moreover, 52% of women who had been diagnosed with preeclampsia and experienced placental abruption have mutations in genes. However, only 15% of women who had been diagnosed with preeclampsia but had not experienced the placental abruption have mutations.

Related Statistics

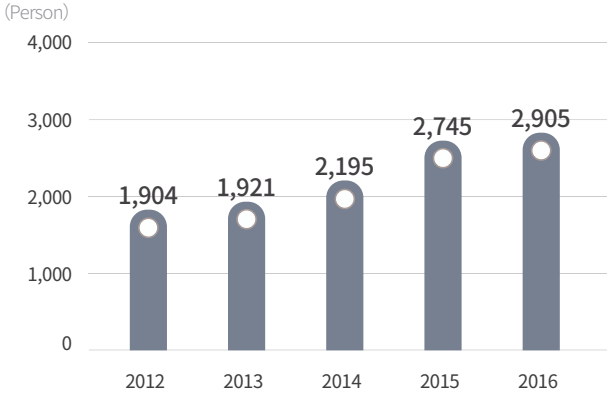
Incidence of Preeclampsia

3~7%

Preeclampsia is a pregnancy-related disease, affecting 3~7% of total pregnancy. It mainly occurs after 20 weeks of pregnancy. One out of 200 preeclampsia patients can have eclampsia with spasm. Therefore, it is recommended to treat that disease as soon as possible.

*Source: 2018. Korean Society for Laboratory Medicine

Number of Preeclampsia Patients by Year



*Source : 2016. Health Insurance Review & Assessment Service

Preeclampsia

How to Prevent Preeclampsia?

Construct Healthy Pregnancy Plan

Advanced maternal age and obesity can increase the risk of preeclampsia and early placental abruption. Therefore, it is very important to consider age prior to pregnancy as well as maintain healthy body condition by proper diet.

Physical Examination by Doctors

Even though there is no convinced treatment for preeclampsia, in case of preeclampsia, it is recommended to have a cesarean section or early labor induction for preventing early placental abruption and other complications. Also, it is important to take a physical examination frequently about diseases. People who are planning for pregnancy should be kept in mind about basic knowledge such as no smoking, taking healthy nutrients before delivery, and taking enough rest. If women who need to control their blood tension or who have diabetes, have to take care of their body. Aspirin can be prescribed to women who had experienced preeclampsia in the previous pregnancy. Calcium can be prescribed to women who are deficient in calcium.

Did You Know?

- Only pregnant women have a risk of preeclampsia.
- The risk of preeclampsia is highest in the first pregnancy.
- During pregnancy, it is best to stop smoking. Because smoking can cause several problems such as preterm birth, early placental abruption, and abnormal growth of a baby. Also, it is not good for both mother and baby.

Preeclampsia

Reference Sites

- **American Pregnancy Association** http://americanpregnancy.org
- **Genetic Home Reference** https://ghr.nlm.nih.gov
- **Healthcare Big-data hub** http://opendata.hira.or.kr
- **Korean Society for Laboratory Medicine** https://labtestsonline.kr
- **Mayo Clinic** http://www.mayoclinic.org
- **MedicineNet.com** http://www.medicinenet.com
- **NAVER encyclopedia** https://terms.naver.com
- **Samsung Seoul hospital** http://www.samsunghospital.com
- **WebMD** http://www.webmd.com

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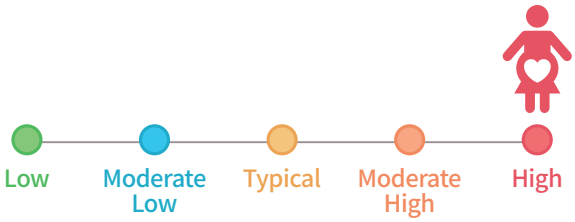
Type 2 Diabetes on Newborn's Weight

Overview

Newborn's weight is generally 2.5kg to 4.0kg. It is affected by not only mother's genetic factors but also environmental factors such as the concentration of insulin in the body. According to some studies, women, who have a specific mutation of type 2 diabetes causing genes, and women who do not have it show different newborn's weight when they give birth. Type 2 diabetes is a metabolic disease when insulin, a hormone to control the level of blood sugar, cannot react properly. Genetic factors in type 2 diabetes are **about 49%**[1] of total causes and the family history of diabetes can also affect that disease. This report provides your genetic information about type 2 diabetes.

Genotyping Results and Risk Assessment



Genetic Risk: **High**
Total Relative Risk: **86%**

Related Gene (Chromosome Location)	My Genotype (Risk Level)	Relative Risk	Genotype Frequency in East Asian
GCK (7p13)	CT Slightly High	1.1	CC Typical 66.79% / CT Slightly High 29.70% / TT High 3.51%
MTNR1B (11q14.3)	GG High	1.2	CC Typical 31.45% / CG Slightly High 50.15% / GG High 18.40%
TCF7L2 (10q25.2)	TT High	1.2	CC Typical 94.70% / CT Slightly High 5.23% / TT High 0.07%

● Risk Allele marked in pink

This report can only provide information on genetic factors so that it does not contain any environmental factors such as exercise habit or living environment. However, these environmental factors can also affect newborn's weight.

1) Sanghera et al. 2012. Type 2 diabetes genetics: beyond GWAS.

Type 2 Diabetes on Newborn's Weight

Related Genes

GCK Gene

GCK gene is one of the blood sugar related genes. An enzyme, produced by *GCK* gene plays very important role in controlling insulin secretion. Reduced insulin secretion can induce diabetes. In case of pregnant woman, the level of blood sugar and energy storage of the fetus can be increased which can affect the weight of newborn. According to studies, a woman with specific genetic type of this gene has increased risk of diabetes. Also, when she is pregnant, the newborn's weight can be increased.

MTNR1B Gene

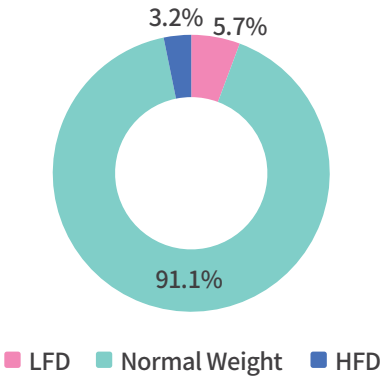
The protein, made by *MTNR1B* gene, binds to melatonin hormone. Melatonin can interact with biorhythm of living organism and affect the secretion of insulin. According to related studies, in case of a specific mutation of *MTNR1B*, insulin secretion cannot function successfully. This can increase the risk of type 2 diabetes.

TCF7L2 Gene

Recent studies show that risk factors of *TCF7L2* gene can influence on type 2 diabetes as well as newborn's weight. A pregnant woman, with a specific allele of this gene, shows a decrease in insulin secretion and an increase in newborn's body weight. If insulin secretion is insufficient, the level of blood sugar is increased. High level of blood sugar in during pregnancy can induce weight gain in a newborn baby.

Standard Weight of Newborn

Rate of Newborn by Weight



*Source: 2015. Health and Welfare Data Portal

Standard weight of a newborn baby is between 2,500g and 4,000g. Half of the newborn's weight is included in 3,000~3,500g. According to the standard of WHO, babies whose weight is lower than 2,500g is classified as light for dates(LFD) and babies whose weight is more than 4000g is classified as heavy for dates(HFD).

- **LFD(Light for dates):** newborn babies whose weight are lower than standard birth weight during gestational age*.
- **HFD(Heavy for dates):** newborn babies whose weight are higher than standard birth weight during gestational age.

*Gestational age: it is same as pregnancy period. It is taken from the woman's last menstrual period

Type 2 Diabetes on Newborn's Weight

Factors to Influence on Newborn's Weight

- **Genetic Factors**

  Newborn's weight can be inherited by parents. However, mother's birth weight can affect on newborn's weight more than father's birth weight.

- **BMI before Pregnancy**

  Women whose BMI is lower than 19 before pregnancy tend to have a baby whose weight is lower than normal weight. Contrast to this case, women whose BMI is high before pregnancy tend to have a baby whose weight is higher than normal weight.

- **Age of Pregnant Women**

  In case of pregnant women over age 35, tend to have a baby with higher weight than a baby of younger women. Also, advanced maternal age pregnancy can increase the risk of several complications, related to pregnancy diseases.

How to Prevent Type 2 Diabetes During Pregnancy?

Exercise

Exercise is very important for managing type 2 diabetes. Regular exercise increases insulin sensitivity which helps reduce blood sugar. In addition, muscle contraction through exercise consumes glucose as an energy source. Therefore, it is effective in lowering blood sugar.

Measure Blood Sugar Regularly

Regular blood glucose measurement is critical to the health of both the mother and a baby. Pregnant women can systematically manage their blood sugar by adjusting a diet and exercise to examine their blood sugar level.

Regular Meal and Healthy Diet

Pregnant women have to eat meals regularly with healthy nutritional intake because their blood sugar is lowered when they skip the meal or eat the meal irregularly. During pregnancy, it is important to maintain carbohydrate and fiber intake. Also, if pregnant women have diabetes, they are encouraged to consume carbohydrates through vegetables, whole grains, and legumes.

Type 2 Diabetes on Newborn's Weight

Reference Sites

- **American Diabetes Association** http://www.diabetes.org
- **Genetic Home Reference** https://ghr.nlm.nih.gov
- **Health and Welfare Data portal** https://data.kihasa.re.kr
- **NAVER encyclopedia** https://terms.naver.com
- **Pregnancy & Baby** http://www.pregnancyandbaby.com
- **What to Expect** http://www.whattoexpect.com

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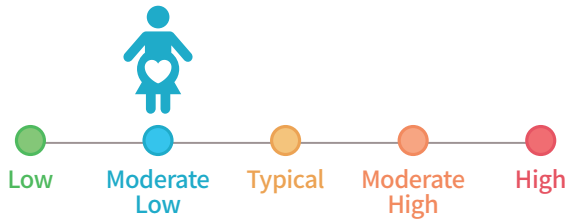
Preterm Birth

Overview

Preterm birth refers to the early birth of a baby before 37 weeks of pregnancy. Preterm birth can increase the risk of diseases in babies after birth. Also, it can be the main cause of high death rate of babies. According to related studies, the death rate of preterm baby is 40 times higher than live birth. Worldwide, 15 million babies are born with preterm birth every year and 1 million babies are dying because of preterm birth complications. Preterm birth can be induced through weakening of cervix, an abnormality of the uterus, experience of preterm birth, infection, malnutrition, drug, age of the mother and other genetic causes. With several studies, *TNF*, *MTHFR*, and *PGR* genes are linked to preterm birth and **30~40% of preterm birth is due to genetic factors**[1].

Genotyping Results and Risk Assessment



Genetic Risk: **Moderate Low**
Total Relative Risk: **40%**

Related Gene (Chromosome Location)	My Genotype (Risk Level)	Relative Risk	Genotype Frequency in East Asian
MTHFR (1p36.22)	**CT** Typical	1.0	CC Typical 32.17% / **CT Typical 50.04%** / TT High 17.79%
PGR (11q22.1)	**AA** Typical	1.0	**AA Typical 89.95%** / AG Slightly High 9.77% / GG High 0.28%
TNF (6p21.33)	**GG** Typical	1.0	**GG Typical 87.78%** / GA Slightly 11.61% / AA High 0.61%

● Risk Allele marked in pink

This report provides information about genetic factors. However, other genetic factors, not involved in this report, or environmental factors can affect the final result. Although it is impossible to change genetic factors, the risk of preterm birth can be reduced by controlling other related factors.

1) Zhang et al. 2017. Genetic associations with gestational length and spontaneous preterm birth.

Preterm Birth

Related Genes

MTHFR Gene

MTHFR(methylenetetrahydrofolate reductase) gene produces an enzyme, related to essential amino acid metabolism. When the activity of this enzyme is decreased, the concentration of homocysteine is increased. Elevated levels of homocysteine in the blood can lead to capillary damage and thrombosis, which decrease blood flow in the placenta, and cause preterm birth.

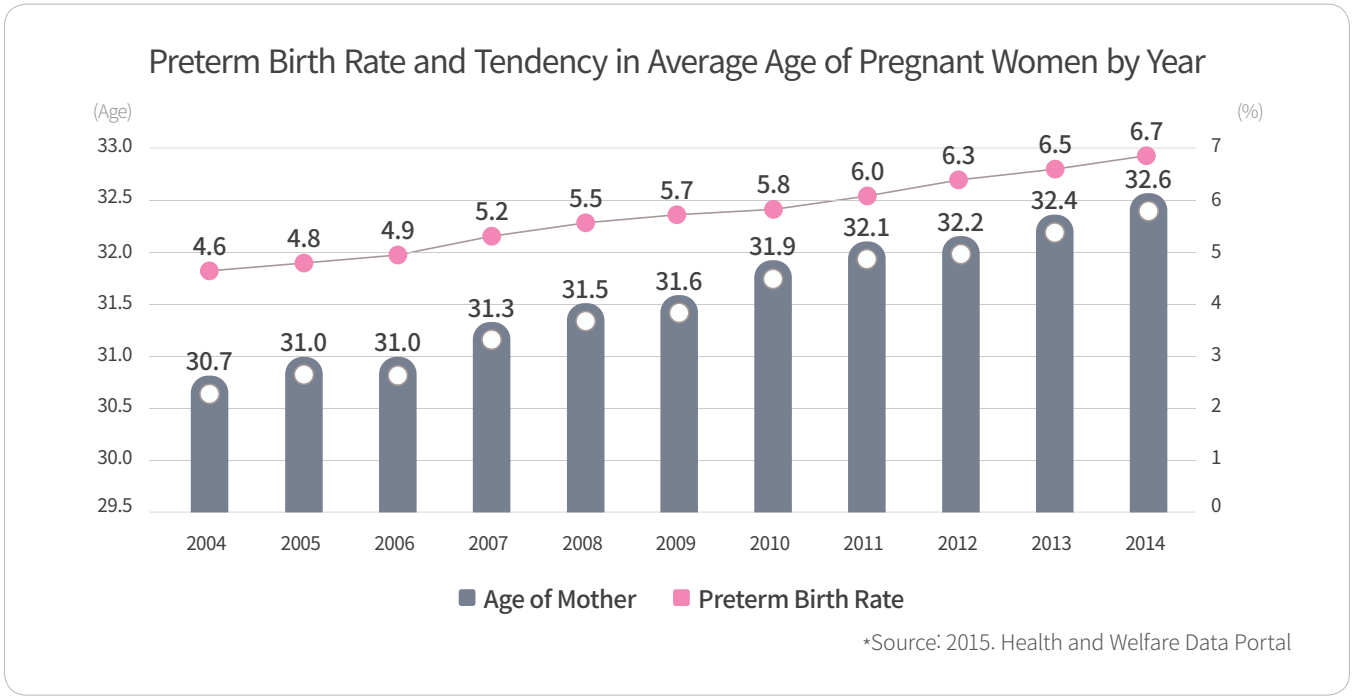
PGR Gene

PGR is the gene to make a receptor of progesterone. Progesterone receptor plays an important role in prevention of preterm birth because it can control the reaction of progesterone, a pregnancy hormone. According to related studies, *PGR* gene mutation is connected to the risk of preterm birth. In case of pregnant women who have a specific mutation in this gene, the risk of preterm birth can increase.

TNF Gene

TNF-α protein, made by *TNF* gene, plays an important role to help the baby to be grown properly. Also, this protein is related to starting and keeping the labor such as breaking of fetal membrane and contraction of the uterus. According to studies, in case of women who have a specific *TNF* genotype, TNF-α production is increased and the pregnancy period can be shorter than normal period.

Related Statistics



*Source: 2015. Health and Welfare Data Portal

Preterm Birth

How to Prevent Preterm Birth?

No Smoking & No Drinking

The components of Tobacco, nicotine and carbon monoxide can interfere with the supply of oxygen to the babies and result in complications like preterm birth. Also, heavy drinking during pregnancy can affect preterm birth. Avoid drinking and smoking for preventing preterm birth.

Healthy Diet

Take sufficient nutrients such as folic acid, calcium, and iron for healthy pregnancy.

Regular Physical Examination

Take a regular physical examination for both mother and her baby's health.

Did You Know?

- If women have a family history of preterm birth or experience of preterm birth, they have a potential risk of preterm birth.
- Pregnant women who have twins or triplets have a high risk of preterm birth.
- In case of Assisted Reproductive Technology, it is possible to have a risk of preterm birth. Because the possibility of having twins can be increased by embryo transfer, it can also increase the risk of preterm birth.

- Preterm birth can happen without symptom. If birth labor starts earlier than expected date, it has to be treated as soon as possible. Through the family history or genetic diagnosis of preterm birth, people can get information about the possibility of preterm birth.

Preterm Birth

Reference Sites

- **Genetic Home Reference** https://ghr.nlm.nih.gov
- **Health and Welfare Date Portal** https://data.kihasa.re.kr
- **Medscape** http://www.medscape.com
- **NICHD** https://www.nichd.nih.gov
- **Science Direct** https://www.sciencedirect.com
- **World Health Organization** http://www.who.int

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