

Premarital counseling and tests



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Please check out this link before viewing the file to know if there are any additions or changes: [medicine Editing](#)

What is premarital counseling ?!

- type of advice that helps couples prepare for marriage.
- can help ensure that both spouses would have a **strong healthy relationship** → giving them a better chance for a **stable and satisfying marriage**.

What is the premarital screening program?!

- In **2004** the Saudi Ministry of Health implemented a **mandatory premarital screening program** of hemoglobinopathies to decrease the incidence of these genetic disorders in future generations.
- In **2008** this test was updated to include **mandatory screening for HBV , HCV and HIV**.
- This new program was named “ **program of healthy marriage**”
(Include hemoglobinopathies genetic disorders +HBV,HCV,HIV)

Pre-requisite of
a screening
program to be
successful:



1) Hemoglobinopathies

Why to include hemoglobinopathies in premarital Screening Program?!

- 1) **Thalassemia & Sickle cell anemia** are fairly **common in** Saudi Arabia , particularly in **eastern and south western regions**
- 2) **high** prevalence of **Carrier status** was reported in the **same regions.**
- 3) **autosomal recessive inheritable** haemoglobinopathies
- 4) These are **incurable** disorders and causes significant morbidity and mortality.
- 5) This imposes a **heavy financial burden** on the society.

How Screening tests can help?!

- A simple blood test can detect **CARRIERS** of these disorders .
- The future couples could be informed about their **chances of producing affected children**



Types of Hemoglobin (analysis of HB electrophoresis results)

NORMAL	
Hb A	<ul style="list-style-type: none"> 92% of adult hemoglobin
Hb A2	<ul style="list-style-type: none"> 2-3% of adult hemoglobin. Increased In βThalassemia.
Hb F	<ul style="list-style-type: none"> less than 1% of hemoglobin in adults. Comprises Normal Hemoglobin in Fetus from 3-9th month of life Increased In β-Thalassemia
ABNORMAL Hemoglobin Chain Production	
Hb H	<ul style="list-style-type: none"> found in α-Thalassemia mild to moderate anemia 2-3 genes are deleted.
Hb Barts	<ul style="list-style-type: none"> found in α-Thalassemia severe form of anemia all 4 genes are deleted. cannot carry oxygen and is incompatible with life infants are stillborn or die immediately after birth (hydropsfetalis).
Hb S Sickle Cell Hemoglobin	<p>presents as Sickle cell Anemia.</p> <ul style="list-style-type: none"> In homozygous state both genes are abnormal (both parent show sickle cell trait) Hb is b/w 6-8 gm /dl.(which is low) Reticulocyte count (immature RBCs):10-20%. Hb electrophoresis : Hb A : 0 % Hb SS :95% Hb F : 2-20% . Sickling Solubility test: precipitation of Hb S gives a turbid appearance .
Hb AS	<p>Sickle cell trait.</p> <ul style="list-style-type: none"> heterozygous state only one chromosome carries the gene effected and one normal gene No symptoms of sickle cell disease but can pass the trait on to their children Hb electrophoresis: Hb A : 60 % Hb SS :40% , Hb F : 2 %
Hb C disease	<ul style="list-style-type: none"> may be associated with Hb S (Hb SC disease) Increased likely hood of thrombosis with life threatening episodes.
Hb E	<ul style="list-style-type: none"> combined defects of Globin chain production and structure. It is combination of β -thalassemia trait and Sickle cell trait . Hb E alone causes mild microcytic anemia .

Male's Dr said (not that important)

A *miscarriage* (sometimes called a spontaneous abortion) is when a baby dies *before* the 20th week of pregnancy. *Stillbirth* is the death of a baby *after* the 20th week of pregnancy

Laboratory Interpretation of Hemoglobinopathies

β -Thalassemia minor (Trait)

symptomless heterozygous carrier state

Normal or slightly low Hemoglobin

↓ mean cell volume (MCV)
And/or
↓ mean cell hemoglobin (MCH)

Hemoglobin A2 Level >3.5%

Microcytic hypochromic picture.

β -Thalassemia Major

severe symptomatic homozygous Anemia

High Hb F & Hb A2

Sickle cell anemia

severe symptomatic homozygous Anemia

high Reticulocyte
Hb A : 0 %
Hb SS :95%
Hb F : 2-20%
Turbid Sickling Solubility test.

may be associated with Hb C → Hb SC disease → thrombosis

Sickle cell trait

symptomless heterozygous carrier state

Hb AS state
(Hb A : 60 %, Hb SS :40%)
Hb F : 2 %

α -Thalassemia

Hb H appearance : mild to moderate
2-3 genes are deleted.

Who is a Genetic Carrier

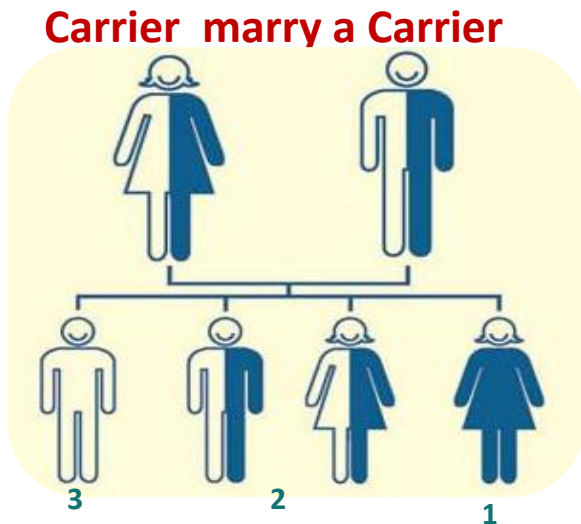
- A person who carries an allele without exhibiting (showing) any effects. (one affected and one normal)
- Such an allele is usually **recessive**, but it may also be **dominant and latent**, with symptoms that do not appear until adulthood.

How will you interpret an Autosomal recessive disorder ?!

- ▶ This disorder manifests itself **only when individual is homozygous** for the disease Allele.
- ▶ The parents are generally unaffected healthy carriers .
- ▶ The offspring of an effected person will be healthy heterozygotes **unless other parent is also a Carrier.**

So symptomatic effected offspring appear only when the two parent are effected or carriers

Possible Future Child's faith: (**important**)



the offspring could be either of the following :

1:homozygous and effected
25% chance (1 in 4 chance)

2: Carrier

50% chance .

3:Genetically Normal
25%chance .

2)STDs

Why to include HIV / HBV /HCV in premarital Screening Program

- ▶ These diseases are now **prevalent in epidemic proportion** .
- ▶ They can be **easily transmitted to sexual partners and to newborns.**
- ▶ They are **not curable** .
- ▶ The mortality and morbidity rates are high.

Who is a viral Carrier ?

One who harbors/carries disease organisms in his body **without manifesting any symptoms**, thus acting as a distributor of infection , Asymptomatic and able to **transmit** disease

A Viral carrier's fate

HIV ,HBV and HCV **CARRIERS** can stay without any symptoms

With early diagnosis and treatment **CARRIERS** of HIV or hepatitis viruses can keep the symptoms under control and reduce the risk of serious complications

Who is HBV Carrier ?

5-10% of patients will not clear the Virus and will become **CARRIERS**

Carriers are usually discovered **incidentally** on blood Test either **Pre marital examination** or **routine health check-up or blood Donation.**

Screening for HCV(ssRNA)

Blood **Transmission** is more common than sexual **Transmission unlike HBV** found incidentally during , Pre marital screening OR routine check-up or Blood donation.

Screening for HIV :

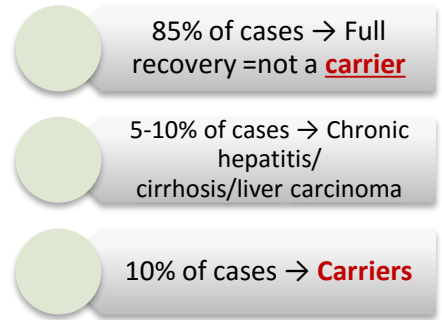
Transmission is sexual --- 60-70% of cases.

From mother to child ---- 90% of cases.

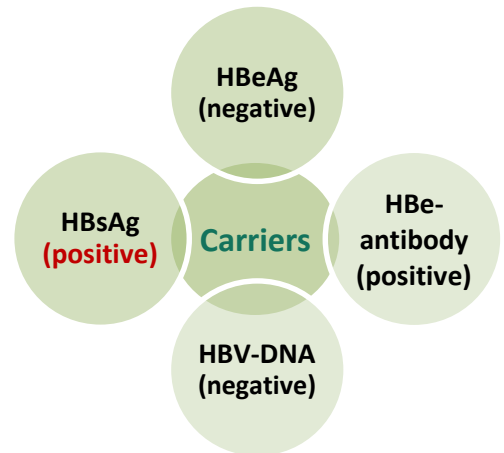
HIV-Antibodies → Confirmed by Western blot Test

The presence of antibody give no indication about the progression of the disease After exposure to HIV –infected person it may take up **to 3 months to become positive** ,⁸ Consider **repeating** this test if exposure may have occurred in less than 3 months prior to testing.

Fate of HBV Infection

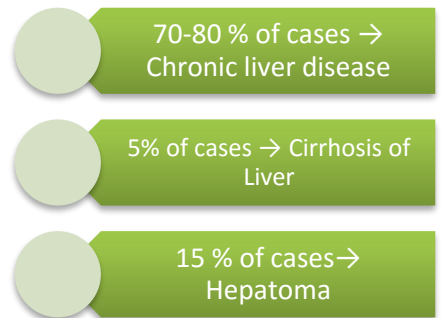


IMPORTANT



Fate of HCV Infection

No carrier state found



What will happen after the tests



Consult your **Family** Physician.

What steps a **Family** Physician should take ?

hemoglobinopathies

- ✓ The future couple should be advised that after marriage your children could suffer from **Sickle Cell anemia or Thalassemia.**
- ✓ The physician **will not issue** the premarital fitness certificate.
- ✓ The decision will be for the future couple whether to go ahead with the marriage or not.

infection with HIV or Hepatitis viruses

- ✓ The physician **will repeat** the test before confirming the diagnosis.
- ✓ If still positive; **will not issue** premarital fitness certificate.
- ✓ **HIV & HCV** Positive are encouraged to avoid **marriage.**
- ✓ In **HBV** Carriers, the healthy partner is advised to be **vaccinated.**
- ✓ The HIV, HCV patient will be informed and referred to a Specialty Clinic for Follow-up.

What Ethical issues can arise :

usually premarital screening comes too **late** for couples to change their **opinions** ABOUT marriage

By this time they are already **committed** for this relationship.

Which Spouse would be affected the Most ?

Male or **Female** ?

Is it a **stigma** or dilemma for female ?

A TABOO FOR FEMALE :

Rejecting marriage on these ground may effect her **Social Life** .

Sometimes this **stigma** may prevent her from **ever getting Married** .

STIGMA FOR MALE or FEMALE :

HIV-testing also has far-reaching social impact especially when someone is planning to marry.

In some communities certain values may clash with concept of premarital HIV-testing with major issues of confidentiality

Available choices after positive Test results :

- ✓ Avoidance of marriage.
- ✓ Those who proceed can be offered reproductive options after prenatal diagnosis.

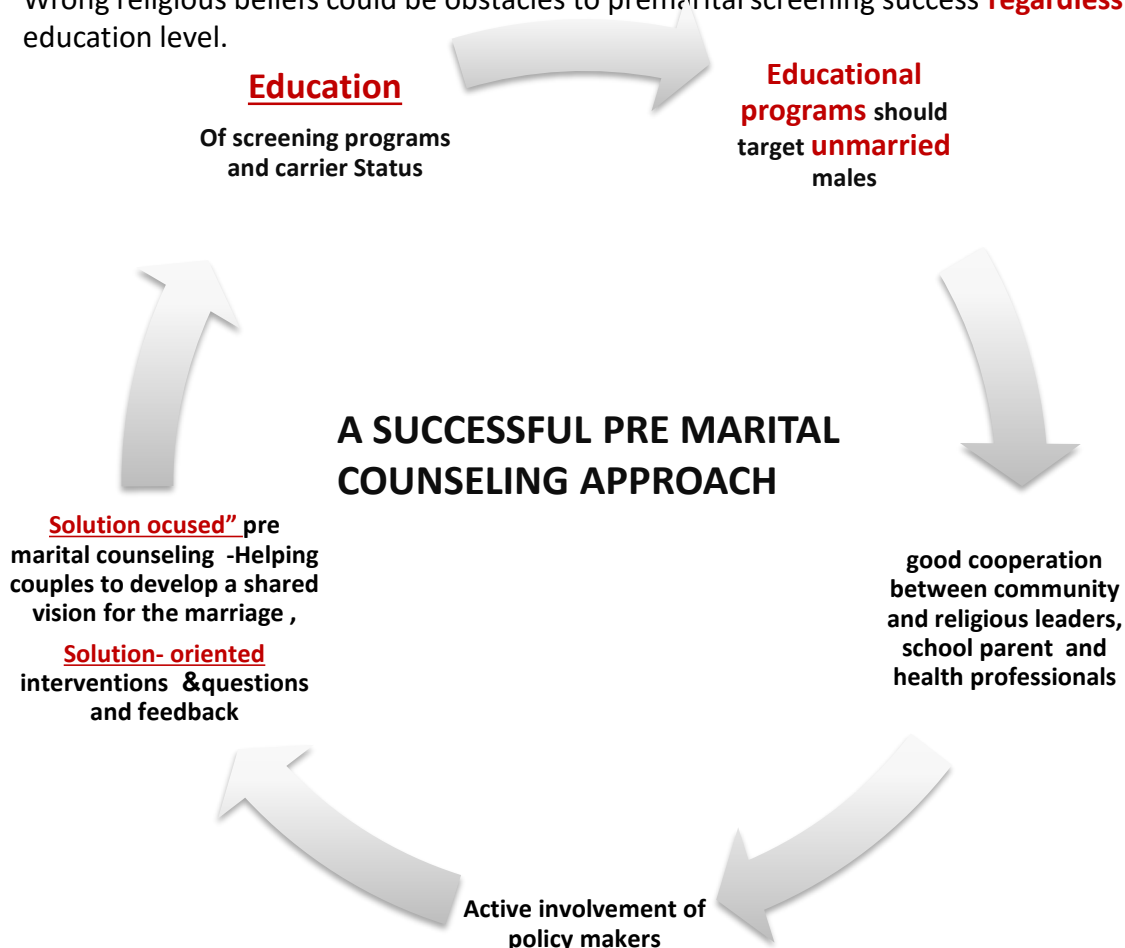
What is the Family Physicians Role:

1:Discuss Genetic counseling :

- ✓ Encourage individual or family to obtain information about a genetic condition that may effect them .
- ✓ so that they can make appropriate decisions about marriage , reproduction and health management.

2:Discuss Consanguinity :

- ✓ Consanguineous marriages → increase the risk for inheriting a recessive allele for a disease.
- ✓ The **closer** the relationship , the greater the risk.(especially cousin marriages)
- **25-60%** of all marriages in Arab regions are **consanguineous**.
- In saudi arabia , 90% of couples detected as **carriers did not follow the advice and went ahead with their marriages**.
- Social and familial commitments make it difficult to ask partners to undergo pre marital testing.
- Wrong religious beliefs could be obstacles to premarital screening success **regardless** of education level.



Summary

Premarital counselling: **is a type of advice** that helps couples prepare for marriage.

Tests performed

- CBC
- Sickle cell test.
- Hemoglobin electrophoresis.
- HBs Ag.
- HBV and HCV screening by ELISA.
- Anti HCV and ANTI HIV
- HIV-Confirmation by Western blot Method.

The program should comply with: ethnic values, social values, economical values ,prevailing culture the test should not be invasive and the sensitivity and specificity should be considered

Hemoglobinopathies: incurable and can cause a potential financial burden, common in eastern and south western regions of Saudi Arabia

STDS like HIV,HBV,HCV :can all be passed to sexual partners and newborns

Sickle cell anemia: in HB electrophoresis

1- HB s appearance:(Sickle Cell Hemoglobin)

HB electrophoresis shows also

- 95% Hb ss
- ↑ HbF
- no HbA
- screening test turbid appearance
- increased reticulocytes

Appearance :

presents as Sickle cell Anemia.

homozygous state both genes are abnormal (both parent of affected child have sickle cell . trait)

When HbC occur with it :called Hb SC disease

2- Hb as carrier

Sickle cell trait.

(heterozygous)only one chromosome is effected

No symptoms of sickle cell disease but can pass the trait on to their children

Hb electrophoresis:

Hb A : 60 % Hb SS :40%

Hb E appear in combination of β - thalassemia triat and Sickle cell trait =mild microcytic anemia

B-thalassemia major: (homozygous)

hbf and hba2 are both increases

b-thalassemia minor (trait): (heterozygous) symptomless

normal to slightly reduced amount of HbA and increases in HA2, microcytic hypo-chromatic anemia

HBV patient serology : HBsAg positive

HBeAg : negative

HBe-antibody : positive

HBV-DNA :negative

No carrier state found in HCV

HIV & HCV Positive are encouraged to avoid marriage.

In HBV Carriers, the healthy partner is advised to be vaccinated.

MOST marriages in Arab regions are consanguineous.

a-thalassemia :

HBH :moderate ,only 2-3 genes are deleted

HB barts (major) :which causes hydrops fetalis, this is where 4 genes are delteded

1-Which one of the following screening tests is NOT included in the “program of healthy marriage “ of Saudi Arabia:

- a-Hemoglobin electrophoresis
- b-Anti-HCV.
- c-HBs Ag
- d-PCR detection of HPV DNA

2-Mohammed is a 27 years old Saudi gentleman who loves his 23 year old cousin, Monira. Mohammed and Monira went to KKUH for pre-marital blood testing. 2 weeks later Dr.Fahad gave them the results, knowing that both of them are carriers of Sickle Cell Anemia. Dr.fahad has told them the probability of giving birth to a sickle cell anemic baby is:

- a-25%
- b-50%
- c-75%
- d-100%

3-According to the previous question what should Dr.fahad further do after telling them the possibilities:

- a-He should repeat the blood test
- b-Courage them to get married.
- c-He should not issue the premarital fitness certificate
- d-Telling the couples that they have no right to go ahead with this marriage.

4-Which of the following is the direct influencer of increased incidence of autosomal recessive disorders in Saudi Arabia :

- a-Low social and healthcare awareness
- b-Consanguineous marriages
- c-Some wrong religious beliefs
- d-All

5-A successful pre-marital counseling approach is considered successful when it provides:

- a-Active involvement of policy makers.
- b-Raising awareness through educational programs.
- c-Targeting the young males and females just before they get married.
- d-Both A&B.

Thanks for checking our work

GOOD LUCK

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