



Premarital Screening

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Lecture for Second year Medical
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Outline

- Facts about premarital screening in KSA
- What's Premarital Screening (Medical tests & Counseling).
- Interpretation of premarital test results.
- Understand the fate of a Carrier !
- Ethical considerations
- Successful premarital Screening



- Genetic disorders particularly Hemoglobinopathies like Thalasemia & Sickle cell anaemia are common in Saudi Arabia , particularly in **eastern and southern region.**
- A high prevalence of **Carrier** status was reported predominantly in the eastern and south western regions of Saudi Arabia.



Facts about pre-marital screening

Started as a part of the Ministry of Health's “**Healthy Marriage Program**”.

Royal Decrees:

- In **2004**, the “pre-marital medical test” which screens for genetic disorders became mandatory for issuing a marriage certificate.
- In **2008**, screening for HIV, HBV, and HCV also became mandatory.
- The test is given at different marriage consultation centers, including KSUMC
- The couple is asked to fill out a form that includes information on name, age, national ID, address and phone number.



وكالة الوزارة للصحة العامة
الوكالة المساعدة للرعاية الصحية الأولية
الإدارة العامة لمكافحة الأمراض الوراثية والمزمنة
برنامج الزواج الصحي





Genetic Carrier

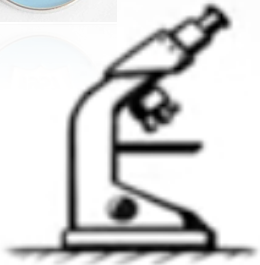
A person who carries or inherited a **recessive allele** for a **genetic** trait or mutation but usually does not display that trait or show symptoms of the disease.

Infectious Dis. Carrier

An infectious **carrier** is a person with inapparent **infection** who is capable of transmitting the pathogen to others. So he is harboring the infection without showing any symptoms.



Benefits of Pre-marital Screening



- A simple blood test can detect **CARRIERS** of genetic or infectious disorders .



- Early prevention of **infectious and genetic diseases**. Helps in providing early intervention to prevent disease

– E.g. :

- Vaccination , Genetic counseling, Nutrition education, Advice regarding contraception, Treatment of infection



- The future couples could be informed about their chances of producing affected child.... Ensuring healthy offspring and healthy community.



- Decrease the economic burden on family ,community & countries



Pre-marital Counseling

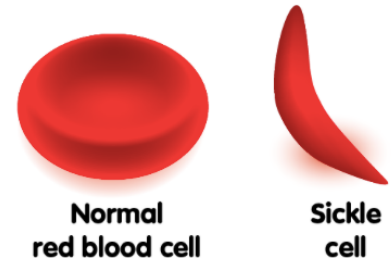
- This is a combination of pre-marital screening tests and subsequent professional health advice on genetic compatibility that is given to a couple prior to marriage.
- The physician advises the couple on risk for contracting the identified infectious disease as well as the risk for having offspring with a hereditary disorder.
- This counseling requires the following:
 - Correct diagnosis of disease or being a carrier.
 - Explanation of the nature of the disease and if treatment is available.
 - Estimation and explanation of genetic risk by drawing a pedigree.
 - Supporting the couple in making the best decision for them.
 - Giving access to long term genetic counseling for incompatible couples .





Pre-marital Screening tests

- This includes the routine tests done before marriage in order to obtain a *pre-marital certificate*, which indicates that the couple are compatible for marriage.
- Routine pre-marital tests are:
 - **Tests for genetic disorders**
 - Sickle cell anemia
 - Thalassemia
 - **Tests for viral infections**
 - Hepatitis B virus (HBV)
 - Hepatitis C virus (HCV)
 - Human Immune Deficiency virus (HIV)





Pre-marital Screening Tests



- CBC
- Sickle cell tests ?
- Hemoglobin electrophoresis?
- HBs Ag.
- HBV serology (ELISA)
- HCV serology (ELISA followed by a RIBA confirmatory test if HCV + ve)
- HIV serology (ELISA) (Anti-HIV , HIV-Confirmation by Western blot Method.



الاسم : الجنسية :
العمر : الجنس : ذكر أنثى
رقم السجل المدني :
العنوان : الهاتف :

HAEMATOTOLOGY RESULT

| TEST | NORMAL RANGE | RESULT | REMARKS |
|---------------------------------|-------------------------------|--------|---------|
| RBC \times L ¹² 10 | M: 4.7 - 6.1 --- F: 4.2 - 5.5 | | |
| HBg/Dl | M: 13 - 18 --- F: 12 - 16 | | |
| Het% | M: 42 - 52% --- F: 37 - 47% | | |
| MCVfl | 80 - 94 | | |
| MCHPg | 27 - 32 | | |
| MCHC/dl | 32 - 36 | | |
| RDW | 11.5 - 14.5% | | |
| Retic | 0.5 - 2 % | | |
| Sikling Test | Positive or Negative | | |
| Hb A | 95 - 97% | | |
| Hb A2 | 2.0 - 3.5% | | |
| Hb F | <1.5% | | |

ABNORMAL HEMOGLOBIN

| HEMOGLOBIN | PATIENT RESULT | HEMOGLOBIN | PATIENT RESULT |
|------------|----------------|------------|----------------|
| Hb S | | Hb J | |
| Hb C | | Hb o-Arab | |
| Hb D | | Gb H | |
| Hb E | | Hb Barts | |
| Hb G | | Hb Barts | |
| Other Hb | | Other Test | |

PREMARITAL SCREENING VIROLOGY TESTS

| | | | |
|-----------------------------|---|---|---|
| HBs Ag Screening by ELISA | <input type="checkbox"/> Positive <input type="checkbox"/> Negative | Anti-HIV & HIV-Ag Screening | <input type="checkbox"/> Positive <input type="checkbox"/> Negative |
| Anti HCV Screening by ELISA | <input type="checkbox"/> Positive <input type="checkbox"/> Negative | HIV Confirmatory by Western Blot Method | <input type="checkbox"/> Positive <input type="checkbox"/> Negative |
| HCV Confirmatory by RIBA | <input type="checkbox"/> Positive <input type="checkbox"/> Negative | HBs Ag Confirmatory | <input type="checkbox"/> Positive <input type="checkbox"/> Negative |

COMMENT :

التوقيع : الطبيب:

ملاحظات هامة :

1. هذه الشهادة تبين نتيجة الفحص المخبري لمرضى الأنيميا المنجلية ، الثلاسيميا ، نقص المناعة المكتسبة والتهاب الكبد الوبائي، ولا تشمل أي أمراض أخرى.
2. هذه الشهادة صالحة لمدة ستة أشهر من تاريخ سحب العينة.

الختم

تقرير الفحص قبل الزواج

| العمر | الجنس | العائلة | الجد | الأب | الاسم |
|---------------|-------|---------|------|------|----------|
| | | | | | |
| العنوان: | | | | | المدينة: |
| الهاتف: | | | | | الجوال: |
| السجل المدني: | | | | | |

فحص الأمراض المعدية: (نقص المناعة المكتسب، التهاب الكبد الوبائي ب/ج):

| | | |
|---------------------------|-----------------------------------|-------------------------------|
| نقص المناعة المكتسب : | <input type="checkbox"/> مصاب | <input type="checkbox"/> سليم |
| التهاب الكبد الوبائي (ب): | <input type="checkbox"/> مصاب | <input type="checkbox"/> سليم |
| التهاب الكبد الوبائي (ج): | <input type="checkbox"/> مصاب | <input type="checkbox"/> سليم |
| اسم الطبيب: | التوقيع: التاريخ: 14 / / هـ | |

فحص الأمراض الوراثية: (الأتيميا المنجلية، والثلاسيميا):

| | | | |
|---|--|--|-------------------------------------|
| <input type="checkbox"/> متوافق | <input type="checkbox"/> غير متوافق بسبب وجود: | <input type="checkbox"/> الأتيميا المنجلية | <input type="checkbox"/> الثلاسيميا |
| Diagnosis: | | | |
| | | | |
| | | | |
| اسم الطبيب: التوقيع: التاريخ: 14 / / هـ | | | |

إقرار للحالات الموجبة:

| |
|---|
| <input type="checkbox"/> أوافق على إبلاغ الطرف الآخر بنتيجة الفحوصات المخبرية الطبية. |
| <input type="checkbox"/> أوافق على إتمام الزواج وشرح لنا طبيعة كافة الفحوصات ونتائجها بعدة المدى. |
| الاسم: التاريخ: 14 / / هـ |
| إقرار ولي أمر المواطنة في حالة إصابة الشاب بأي من الأمراض الوراثية أو المعدية. |
| اسم الوالي: التاريخ: 14 / / هـ |

المرفقات:

نتائج الفحوصات الطبية المخبرية. صورة من شهادة الفحص قبل الزواج



Kingdom of Saudi Arabia
Ministry of Health

المملكة العربية السعودية
وزارة الصحة
مديرية الشؤون الصحية
المحافظة / المنطقة
مستشفى / مركز

الرمز/الرقم:
التاريخ: 14 / / هـ

شهادة الفحص قبل الزواج

تم إجراء الفحص المخبري لأمراض الدم الوراثية (الأنيميا المنجلية، والتلاسيميا) والأمراض المعدية (نقص المناعة المكتسب "الإيدز" والالتهاب الكبدي (ب، ج) لكل من:

| الاسم | الأب | الجد | العائلة |
|---------------|---------|--------|---------|
| السجل المدني: | | | |
| رقم الشهادة | تاريخها | مكانها | |

و

| الاسم | الأب | الجد | العائلة |
|---------------|---------|--------|---------|
| السجل المدني: | | | |
| رقم الشهادة | تاريخها | مكانها | |

- ملحوظات: (إن مدة صلاحية الفحص الطبي بالنسبة للأمراض المعدية 6 أشهر فقط)
- 1- فحوصات الأمراض المعدية تعبر لنتائجها المخبرية عن نتائجها الموجبة أو السلبية حتى تاريخ أخذ العينة.
 - 2- تم شرح آثار حالات الزواج غير المتوافق وراثياً لطرفي العقد.
 - 3- يعاد فحص الأمراض المعدية مع كل عقد نكاح.

اسم طبيب عيادة المشورة
التاريخ 14 / / هـ
التوقيع

اسم مدير المستشفى
التوقيع

الختم الرسمي



Why to include hemoglobinopathies in premarital Screening Program

- These are **autosomal recessive** inheritable haemoglobinopathies .
- Common in some regions of Saudi Arabia.
- These are **incurable disorders** and causes significant morbidity and mortality.
- This imposes a heavy financial burden on the society.



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

- These diseases are now prevalent in epidemic proportion .
- They can be easily transmitted to the partner & to newborns.
- They are not curable .



Types of Normal Hemoglobin

- **1. Hb A**

comprises 92% of adult hemoglobin.

- **2. Hb A₂**

Comprises 2-3% of adult hemoglobin. **Increased In β -Thalassemia.**

- **3. Hb F**

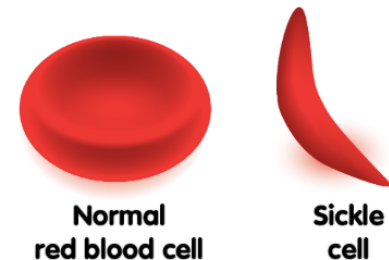
Comprises less than 1% of hemoglobin in adults. Normal Hemoglobin in Fetus from 3-9th month of life. **Increased In β -Thalassemia.**



Pre-marital Screening Tests

Sickle cell test:

- This is formed to detect people who have abnormal Hb in their blood
- A positive sickle cell test tells us the person has abnormal Hb, which can either be **sickle cell trait** or **sickle cell disease**. It CANNOT differentiate between those two.
- If positive, one must do electrophoreses and further genetic testing to identify type of hemoglobinopathy and severity.





Examples of Abnormal Results for Genetic Disorders

Sickle Cell Trait

- When a person is heterozygous for the sickle cell gene. (inherits one sickle cell gene)
- It normally doesn't cause symptoms of anemia
- These people have a small percentage of HbS. Hb electrophoresis Shows = Hb A : 60 % , Hb SS :40% , Hb F : 2 % .
- **This person is a *carrier*** and can transfer the gene to offspring.

Sickle cell disease

- When a person is homozygous for the sickle cell gene (inherits the sickle cell gene from each parent)
- These people have a large percentage of HbS
- HbS >80%, while HbA₂ 2-4.5% and HbF 1-20%
- These people **express** the disease and can transfer the gene to offspring.



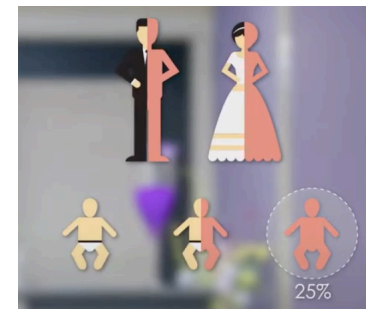
Examples of Abnormal Results for Genetic Disorders

α Thalassemia Carrier

- This person has normal or slightly low Hb
- They usually don't express symptoms of anemia
- Low MCV, Low MCH, HbA₂ is >3.5% by electrophoresis
- These people are **missing** one alpha-globin protein gene
- This person can transmit the altered gene to offspring, and if marries another person with the same trait, their child has 25% chance of having Thalassemia

β Thalassemia minor (Trait)

- This person usually Symptomless
- There is one altered gene for beta-globin protein (heterozygous carrier state).
- This person can transmit the altered gene to offspring, and if marries another person with the same trait, their child has 25% chance of having Thalassemia





How will you interpret an Autosomal recessive disorder

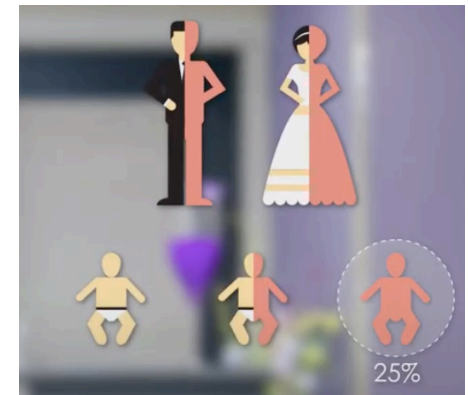
- This disorder manifests itself only when individual is homozygous for the disease Allele.
- The parents are generally unaffected healthy **carriers** .
- The offspring of an effected person will be healthy heterozygotes **unless other parent is also a Carrier.**



Possible Future Child's fate

So when **Carrier** marry a **Carrier** ; the **offspring** could be either of the following :

- **homozygous and affected --25% chance (1 in 4 chance)**
- **A Carrier ----- 50% chance .**
- **Genetically Normal ----- 25% chance .**





A Viral carrier's fate

- HIV and Hepatitis B & C viruses can remain dormant for months or even years in **CARRIERS** without showing 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.



Examples of abnormal results for Viral infection testing

When HB-sAg tests positive, one must do a confirmatory test

HBV chronic infection

- HB-sAg positive
- Anti-HBc positive
- Anti-Hbs negative

HCV

- If anti-HCV screening is positive, supplemental anti-HCV (RIBA) should be done for confirmation.

HIV

- If a person is positive for Anti-HIV and HIV-Ag & Western Blot, then that person is considered infected.
- It is best to **repeat test before confirming diagnosis**



Interpretation of results

The results of premarital screening will let us know whether the couple is *compatible* or *incompatible*.

Compatible

- When neither one of the couple is a carrier for sickle cell disease or β -thalassemia, or only one of the two is a carrier.



Incompatible

- When both members are carriers for either or both sickle cell disease and β -thalassemia. Or when one has the disease and the other is a carrier.



Interpretation of results cont.

- Results should be disclosed to each person in an ethical matter.
- After results are disclosed, it is *totally their decision* whether or not to go through with the marriage and it should be respected.
- In case the couple are incompatible, the physician should clearly explain the nature and prognosis of the inherited disease as well as the genetic risk for offspring.
- Remember that routine pre-marital screening only screens for sickle cell and thalassemia. Consanguineous couples should be advised about other probable genetic disorders found in their families.
- If a person is positive for **HIV or hepatitis**, they will be referred to a specialist for treatment and they **WILL NOT** be issued a pre-marital certificate.



Premarital Screening (Counseling Clinics)



Pre-implantation Genetic Diagnosis (PGD)

- In case the **incompatible** couple chooses to go through with the marriage, they will have the option of testing for the genetic disorder in their offspring prior to implantation of the embryo in the uterus.
- **PGD** is a technique in which embryos of IVF can be tested for genetic disorders.
- It is advised when a couple's offspring may be at serious risk of an inherited genetic disorder.
- **Candidates for procedure:**
 - Parents who previously had a child with a genetic disorder
 - A couple with repeated pregnancy losses due to genetic disorder
 - A couple at high risk of having a child with a genetic disorder



In case of 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(for now) – as there is much higher chance to transmit infection to your future spouse.
- 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.



Ethical Considerations

- Many couples decide to go through with the marriage **despite of incompatibility**
- It has been heavily debated on whether or not to forcefully prevent incompatible marriages. **However, there is no law to prevent it.**
- Pre-natal diagnosis could lead to termination of pregnancy and emotional distress. By law, termination of pregnancy is only allowed in the first 120 days after conception.

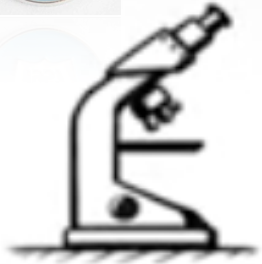


Successful Pre-marital Screening

- Pre-marital screening benefits can only be obtained when there is **strict** implementation of the “Pre-marital Screening or healthy marriage Program”.
- Public education about genetic disorders and how they are inherited is very important.
- Education about modes of transmission of HIV, HBV and HCV and how it can be prevented is also crucial.
- Pre-marital screening success depends on the society’s attitude and beliefs towards pre-marital screening program should be addressed and misconceptions should be corrected.



Benefits of Pre-marital Screening



- Early disease detection
 - Early prevention of infectious and genetic diseases.
- Helps in providing early intervention to prevent disease

– E.g. :

- Vaccination in case of HBV +ve partner
- Genetic counseling
- Nutrition education
- Advice regarding contraception
- Treatment of infection



Decreases risk factors for genetically determined illnesses and improves the quality of life for couple and offspring. Ensuring healthy offspring and healthy community.



- Decrease the economic burden on family ,community & countries
- Improves communication skills and overall couple relationship



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Good luck !

Reference & further information:

MOH : <https://www.moh.gov.sa/en/HealthAwareness/Beforemarriage/Pages/default.aspx>
<https://www.moh.gov.sa/Ministry/About/Health%20Policies/004.pdf>

For any inquiry:
FCM@KSU.EDU.SA

Courtesy to :
Family medicine department , KSU.