MEDICINE

Premarital Counselling And Tests

OBJECTIVES :

- What is premarital counseling.
- Who are Carrier's and their fates.
- How to interpret the TESTS.
- What is a successful counseling.

مصدر شامل لمحاضرة البنات والأولاد

Revised by
خولة العماري & هشام الغفيلي

FOR FURTHER EXPLANATION, CORRECTION OR IF YOU WANT THIS LECTUER IN OTHER FORMAT CONTACT US ON
medicine435.17@gmail.com

By: Wadha AlOtaibi & Amjad Alduhaish

IMPORTANT NOTES EXTRA EDITING FILE

❖ What is premarital counseling :

Premarital counseling is a type of family therapy (or advice) that helps couples prepare for marriage. Premarital counseling can help ensure that both spouses (you and your partner) would have a strong, healthy relationship in addition to giving them a better chance for a stable and satisfying marriage.

▪ What is the pre marital screening program ?

Back ground :

- Genetic disorders **particularly Hemoglobinopathies** like Thalasemia & Sickle cell anaemia are fairly **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.
- In 2004 the Saudi Ministry of Health implemented a mandatory premarital screening program to decrease the incidence of these genetic disorders in future generations. initially the test was focused on the genetic part.
- In 2008 this test was updated to include mandatory screening for HBV , HCV and HIV. they included the STD part.(sexually transmitted diseases)
- This new program was named "program of healthy marriage".
- Its valid only for 6 months.

▪ What are the TESTS Performed ?

1. Complete Blood Count.
2. Sickle cell test.
3. Hemoglobin electrophoresis.
4. HBs Ag.(hepatitis B surface antigen)
5. Anti-HCV.
6. HBV screening by ELISA..
7. HCV screening by ELISA.
8. Anti-HIV.
9. HIV-Confirmation by Western blot Method.

▪ Pre-requisite of a screening program :

Any succussesful screening program must comply with;

1. Prevailing culture.
2. Ethnic values.
3. Economic values .
4. Societal values.

(つ ´ ω`)ﾉ(╥ω╥) more 7 pages you can do it !

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.

How Screening tests can help ?

- A simple blood test can detect CARRIERS of these disorders .They don't do molecular studies, only electrophoreses so we won't know what gene is exactly affected, we will only know if they are affected or carries !
- The future couples could be informed about their chances of producing affected children. Also educate them about possible solutions such IVF if "for example" they have high risk for sickle cell anemia like if he's affected and she's a carrier or BOTH are carriers & they choose to take the risk.

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 new borns.
- They are not curable .
- The mortality and morbidity rates are high.

Laboratory Interpretation of Hemoglobinopathies :

1. β -Thalassemia minor (Trait): symptomless heterozygous carrier state.
2. β -Thalassemia Major: severe symptomatic homozygous Anemia.
3. Sickle cell anemia.
4. Sickle cell trait.

Sometimes trait has symptoms like in sickle cell trait male could have painful erection.

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.

Obviously filling an empty space ☺

(っ ´ ω`)ﾉ(╥ω╥) more 6 pages you can do it !

▪ Types of Abnormal Hemoglobin Chain Production :

1. **Hb H** -- found in α-Thalesemia.

✧ It is mild to moderate anemia, when 2-3 genes are deleted.

2. **Hb Barts** -- found in α-Thalesemia.

✧ It is severe form of anemia, when all 4 genes are deleted.
✧ Cannot carry oxygen and is incompatible with life.
✧ Infants are still born or die immediately after birth (hydropsfetalis).

3. **Hb S** -- Sickle Cell Hemoglobin.

✧ In homozygous state both genes are abnormal – presents as Sickle cell Anemia.
Hb is b/w 6-8 gm /dl.
Reticulocyte count is 10-20%.
Hb electrophoresis Shows = Hb A : 0 % ,
Hb SS :95% ,
Hb F : 2-20% .
✧ Sickling Solublity test: precipitation of Hb S gives a turbid appearance .
The parents of affected child will show sickle cell trait.

4. **Hb AS** -- Sickle cell trait.

✧ In heterozygous state only one chromosome carries the gene.
Hb electrophoresis Shows = Hb A : 60 % ,
Hb SS :40% ,
Hb F : 2 % .

5. **Hb C disease** -- may be associated with Hb S (Hb SC disease)

✧ Increased likely hood of thrombosis with life threatening episodes.

6. **Hb E** -- combined defects of Globin chain production and structure.

✧ **It is combination of β -thalassemia triat and Sickle cell trait .**
✧ Hb E alone causes mild microcytic anemia .

(っ´ω`)ﾉ(╥ω╥) more 5 pages you can do it !

❖ Who are Carrier's and their fates ?

▪ Genetic Carrier :

A person who carries an allele without exhibiting its effects. Such an allele is usually recessive, but it may also be dominant and latent, with symptoms that do not appear until adulthood.

▪ Who is a carrier of Thalassemia ? (Asymptomatic)

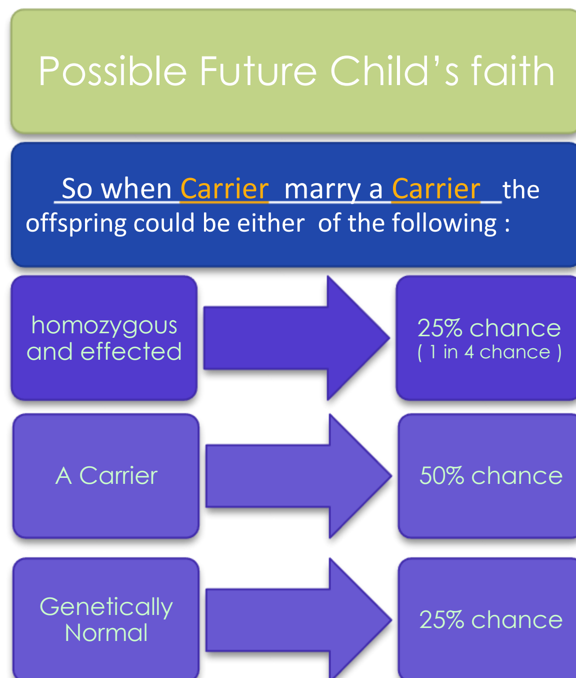
The β Thalassemia Trait is indicated by the following;

1. Normal or slightly low Hemoglobin.
2. Decreased mean cell volume (MCV) (MCV=the average volume of a red blood cells)
3. And/or reduced mean cell hemoglobin (MCH).
4. Hemoglobin A2 Level >3.5% by Hemoglobin electrophoresis.
5. Microcytic hypochromic picture. The problem here is that patients go to private hospitals and are given iron supplements, because they will see the picture as iron deficiency anemia and usually they don't check what is behind it (could be thalassemia or sickle cell anemia) this will cause iron toxicity to the patient, the solution is to always include ferritin in iron deficiency investigations. Or to do hemoglobin electrophoresis.

Read about ferritin test here

▪ 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.



Every single pregnancy has :
25% chance of an affected child
25% chance of a normal child
50% chance of a carrier child

So, they could have all their offspring normal or carriers or affected!

We should explain to the couple that these percentages do not indicate that 25% of their offspring will be affected and 50% will be carrier and only 25% will be normal !! This is always a misunderstanding !!

(つ´ω`)ﾉ(╥ω╥) more 4 pages you can do it !

Name of carrier	Who is it ?	Its fate..
Viral Carrier	One who harbors disease organisms in his body without manifesting any symptoms, thus acting as a **distributor** of infection.	✧ 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.
HBV – Infection	Following an acute HBV infection , which may be sub-clinical 5-10% of patients will not clear the Virus and will become **carrier's** of HbsAg. **Carriers** are usually discovered incidentally on blood Test either Pre marital examination or routine health check-up or blood Donation.	✧ 85% of cases → Full recovery ✧ 5-10% of cases → Chronic hepatitis/cirrhosis/liver carcinoma ✧ 10% of cases → Carriers. Screening for HBV (double stranded DNA –Virus)
HCV – Infection	---	**No carrier state found** ✧ Chronic liver disease → 70-80 % of cases. ✧ Cirrhosis of Liver → 5% of cases. ✧ Hepatoma → 15 % of cases
HIV- Antibodies	---	✧ Confirmed by Western blot Test. ✧ Presence of HIV-antibodies gives no indication about disease progression. After exposure to HIV –infected person it may take up to 3months to become positive. ✧ Consider repeating this test if exposure may have occurred < than 3 months prior to testing.

(っ´ω`)ﾉ(╥ω╥) more 3 pages you can do it !

Healthy HBsAg Carriers :

- HBsAg : positive.
- HBeAg : negative.
- HBe-antibody : positive.
- HBV-DNA : Negative.

Screening for HCV :

- A single stranded RNA Virus.
- It is 70-90 % of cases found in post-transfusion cases.
- Again mostly found incidentally during Pre marital screening OR routine check-up or Blood donation.
- Not easily spread through sexual – contact.

Screening for HIV :

- HIV is a Retrovirus infecting T-Helper cells bearing the CD4 receptors.
- Transmission is sexual → 60-70% of cases.
- From mother to child → 90% of cases.

What will happen after the tests ?

- Consult your Family Physician

What steps a Family Physician should take ?

In case of carrier for 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.

In case of infection with HIV or Hepatits 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.

(っ ´ ω`)ﾉ(╥ω╥) more 2 pages you can do it !

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.

A TABOO for FEMALE :(TABOO= practice that is prohibited or restricted by social or religious custom)

- 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.

What is the Family Physicians Role ?

A. 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.

B. Discuss Consanguinity :

- Relationships by blood or common ancestry, in which the chances of offspring inheriting a recessive allele for a disease are increased .
- The closer the relationship , the greater the risk.(especially cousin marriages)

Prevalence of Consanguineous marriages in Muslim Community:

- 25-60% of all marriages in Arab regions are consanguineous , with a high incidence of first-cousin marriage.
- In Saudi Arabia , 90% of couples detected as carriers did not follow the advice and went ahead with their marriages.
- There are many teachings in Islamic Culture which promote healthy marriage and role of counseling.

Marriages between members of same tribe or extended family groups are favored in Muslim communities. Social and familial commitments make it difficult to ask partners to undergo premarital testing. Wrong religious beliefs could be obstacles to premarital screening success regardless of education level.

❖ What is a successful counseling.

- A SUCCESSFUL PRE MARITAL COUNSELING APPROACH:

Education and attitude of the couples to be screened .

The meaning of the term “carrier Status” should be made known to the members of the public long before they get married.

Educational programs about the benefits of premarital examination should target unmarried males, so they can make informed choices about unmarried females and consanguineous marriages.

Active involvement of policy makers to establish and implement appropriate screening techniques and policies.

“Solution focused” pre marital counseling - Helping couples to develop a shared vision for the marriage.

- Solution- oriented interventions .

- Solution -oriented questions and feedback.

Approach adopted by the counselor . Educate all members of the screening Team (lab technologist ; nurse practitioners; physicians; counselors; out-reach workers ; social workers.)

There should be good cooperation between community and religious leaders, school parent and health professionals.

- Available choices after positive Test results :
  - Avoidance of marriage.
  - Those who proceed can be offered reproductive options after prenatal diagnosis.

- CONCLUSION :

Any mandatory screening program does have the potential to succeed as long as the TARGET POPULATION is clearly identified and all ethical issues (confidentiality of results) ,religious , cultural and human rights and concerns about post-diagnostic management are fully addressed.

(つ ´ ω`)ノ(╥ω╥) The End !