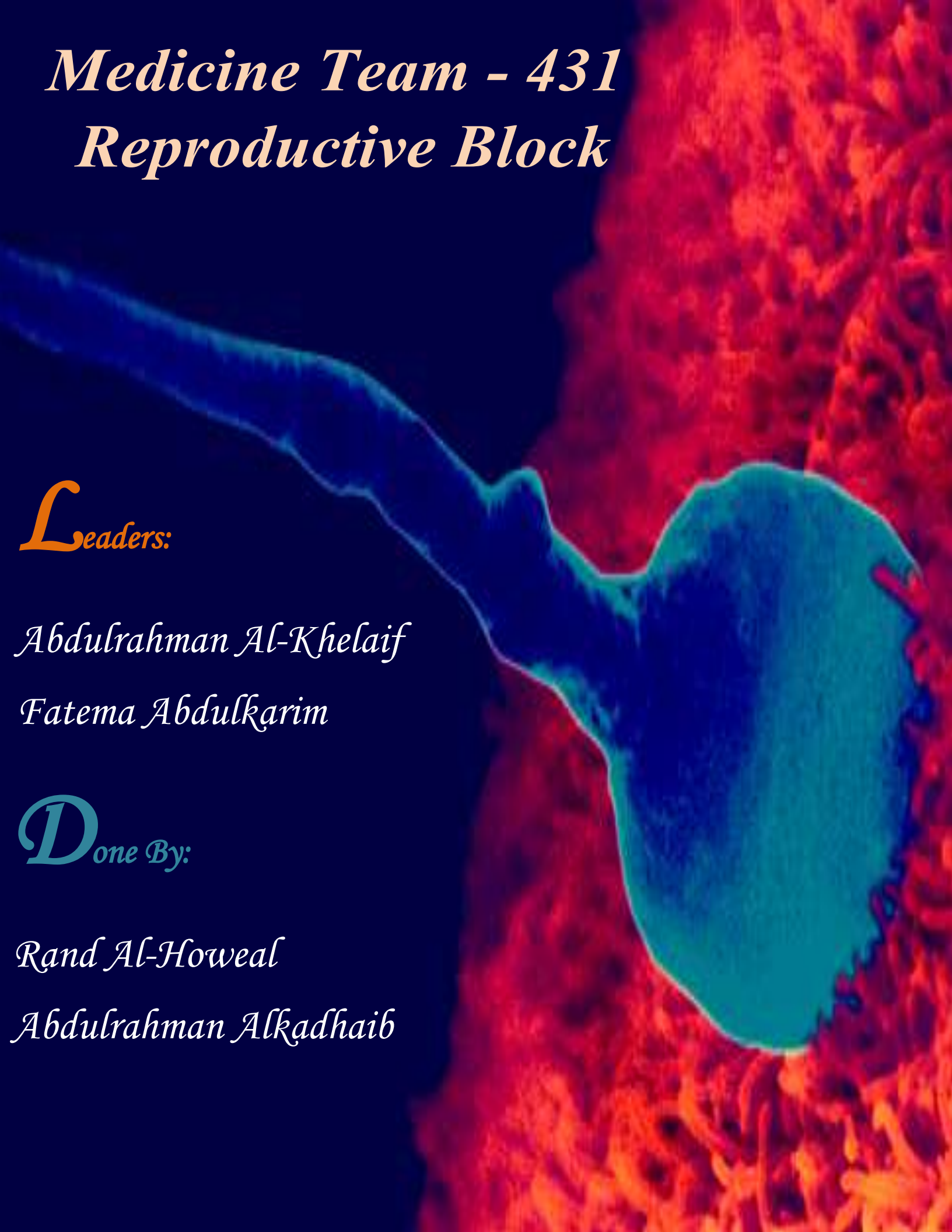
Medicine Team - 431
Reproductive Block



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PRE MARITAL COUNSELING & TESTS

Objectives:

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

What is premarital counseling?

Premarital counseling is a type of advice that helps couples prepare for marriage. Premarital counseling 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 pre marital screening program?

Background:

- **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.**
- **In 2008 this test was updated to include mandatory screening for HBV, HCV and HIV.**
- **This new program was named "program of healthy marriage".**


Kingdom of Saudi Arabia
Ministry of Higher Education
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الفحص المخبري ما قبل الزواج

HAEMATOLOGY RESULT

TEST	NORMAL RANGE	RESULT	REMARKS

PREMARITAL SCREENING VIROLOGY TESTS

COMMENT :

What are the TESTS Performed?

- **Complete Blood Count.**
- **Sickle cell test.**
- **Hemoglobin electrophoresis.**
- **HBs Ag.**
- **Anti-HCV.**
- **HBV screening by ELISA.**
- **HCV screening by ELISA.**
- **Anti-HIV.**
- **HIV-Confirmation by Western blot Method.**

Pre-requisite of a screening program:

Any successful screening program must comply with:

- **prevailing culture**
- **Ethnic values.**
- **Economic values. should not be expensive**
- **Societal values.**

Also in screening programs, you should consider:

- How sensitive and specific the program is.
- It should not be invasive.

Why to include hemoglobinopathies in premarital Screening Program?

- **These are autosomal recessive inheritable haemoglobinopathies which are 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.**
- **The future couples could be informed about their chances of producing affected children**

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.**

Laboratory Interpretation of Hemoglobinopathies:

- **β -Thalassemia minor (Trait): symptomless heterozygous carrier state.**
- **β -Thalassemia major: severe symptomatic homozygous Anemia.**
- **Sickle cell anemia.**
- **Sickle cell trait.**

Types of Normal Hemoglobin:

Hb A	comprises 92% of adult hemoglobin
Hb A2	Comprises 2-3% of adult hemoglobin. Increased In β-Thalassemia
Hb F	Comprises less than 1% of hemoglobin in adults. Normal Hemoglobin in Fetus from 3-9th month of life. Increased In β-Thalassemia

Types of Abnormal Hemoglobin Chain Production:

Hb H	*found in α-Thalasemia*	It is mild to moderate anemia, when 2-3 genes are deleted.
Hb Barts	*found in α-Thalasemia*	It is severe form of anemia , when all 4 genes are deleted. Hb Barts cannot carry oxygen and is incompatible with life. Infants are still born or die immediately after birth (hydrops fetalis).

Types of Abnormal Hemoglobin Chain Structure:

Hb S → Sickle Cell Hemoglobin.

In homozygous state both genes are abnormal – presents as Sickle cell Anemia.

- **Hb is between 6-8 gm /dl.**
- **Reticulocyte count is 10-20%.**
- **Hb electrophoresis Shows = Hb A : 0 % ,**

  Hb SS: 95% ,

  Hb F : 2-20% . __Know that it is high

- **Sickling Solublity test: precipitation of Hb S gives a turbid appearance.**

The parents of affected child will show sickle cell trait.

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 % .

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

Increased likely hood of thrombosis with life threatening episodes.

Hb E → combined defects of Globin chain production and structure.

It is combination of β -thalesemia triat and Sickle cell trait .

Hb E alone causes mild microcytic anemia.

Who is a viral Carrier?

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

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

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.**

Who is a carrier of Thalessaemia

The β Thalassemia Trait is indicated by the following :

* **Normal or slightly low Hemoglobin.**
* **Decreased mean cell volume (MCV)**
* **And/or reduced mean cell hemoglobin (MCH).**
* **Hemoglobin A2 Level > 3.5% by Hemoglobin electrophoresis.**
* **Microcytic hypochromic picture.**

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.**

Interpretation of an Autosomal recessive disorder

- **So when Carrier marry a Carrier ; the offspring could be either of the following :**
- **homozygous and effected --25% chance (1 in 4 chance)**
- **A Carrier ------ 50% chance.**
- **Genetically Normal ------ 25% chance.**

Fate of HBV –Infection

- **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)

Who is HBV Carrier?

- **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.**

Healthy HBsAg Carriers

- **HBsAg : positive. (Marker of infection)**
- **HBeAg : negative. (the patient is non-infectious)**
- **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.**

Fate of HCV –Infection

No carrier state found

Chronic liver disease	**50 % of cases**
Cirrhosis of Liver	**5% of cases**
Hepatoma	**15 % of cases**

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.**

FATE OF 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 3 months to become positive.
- **Consider repeating this test if exposure may have occurred < than 3 months prior to testing.**

What will happen after the tests ?

Consult your Family Physician

What steps a Family Physician should take ?

In case of carrier for hemoglobinipathies:

- **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 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.**

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.**

What is Genetic counseling

- **It is a process by which an individual or family obtains information about a genetic condition that may affect them, so that they can make appropriate decisions about marriage, reproduction and health management.**

What is Consanguinity?

- **This is a 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.**

Prevalence of Consanguineous marriages in Non-Muslim Community

- **Marriage between first cousins is forbidden by the Orthodox Church and Roman Catholic Church.**
- **According to one study the support from CHURCH was the main reason for the success of screening programmes in Cyprus and Greece.**

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 pre marital testing.**
- **Some people believe that their FATE is determined by God and therefore accept the risk of having sick child.**
- **Wrong religious beliefs could be obstacles to premarital screening success regardless of education level.**

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. Like 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.**

Questions:

1- The definition of Genetic Carrier is:

A. A person who carries an allele and exhibits its effects.
B. A person harbors disease organisms in his body without manifesting any symptoms
C. A person who carries an allele without exhibiting its effects.
D. A person who harbors disease organisms in his body and manifests symptoms.

2- The β Thalassemia Trait is indicated by:

A. Increased mean cell hemoglobin (MCH).

B. Decreased mean cell volume (MCV)

C. Hemoglobin A2 Level < 3.5% by Hemoglobin electrophoresis.

D. Macrocytic hyperchromic picture.

3- If both parents are carriers of an autosomal recessive disorder, the chance of having a homozygous affected offspring will be:

A. 100%

B. 75%

C. 25%

D. 50%

Answers: 1- **C** 2- **B** 3- **C**