Pre-marital Screening Clinical Practice Guidelines

Definition

The healthy marriage: A state of wholeness and harmony (physical, psychological, social, and health) between the marriage partners that leads to interaction between several influential factors, inherited and acquired, which are transferred to the children, with the aim of building a sound, stable family successfully performing its role, and fulfilling one of the fundamental human rights.

Premarital screening: Carrying out laboratory tests for partners planning to get married to check for the presence of selected hereditary blood disorders (hemoglobinopathies, especially sickle cell anemia and beta thalassemia) and infectious diseases (hepatitis B and C, and HIV) with the purpose to provide ithe engaged couple with medical counseling on the risk of transmission of those diseases to the spouse or future children and choices and alternatives to help them plan for a healthy family.

Hereditary blood disorders: A group of blood disorders transmitted from parents to children that result from a defect in the structure and components of red blood cells making them unable to carry out their natural function and leading to the emergence of symptoms in affected persons. The following blood disorders are included in the program: sickle cell anemia and beta thalassemia.

Unaffected person: A person who does not carry the disease trait.

Carrier: A person carrying the disease trait but is asymptomatic.

Affected person: A person who carries the disease trait and shows disease symptoms.

Safe marriage (compatible): Any marriage in which one, or both, partner is unaffected by the hereditary blood disorders included in the program, irrespective of whether the other partner is a carrier for the disorder or affected by it.

Unsafe marriage (incompatible): Any marriage in which both partners are affected by a hereditary blood disorder included in the program or are carriers for it, or where one partner is affected and the other is a carrier for a disorder.

Infectious Diseases: Diseases that infect humans through the transmission of viruses from one person to another. The following infectious diseases are included in the program: hepatitis B, hepatitis C, and HIV.

Consultation: A meeting to engage in face-to-face discussion, where one person is able to help another person, couple, family, or group to define their needs and make decisions.

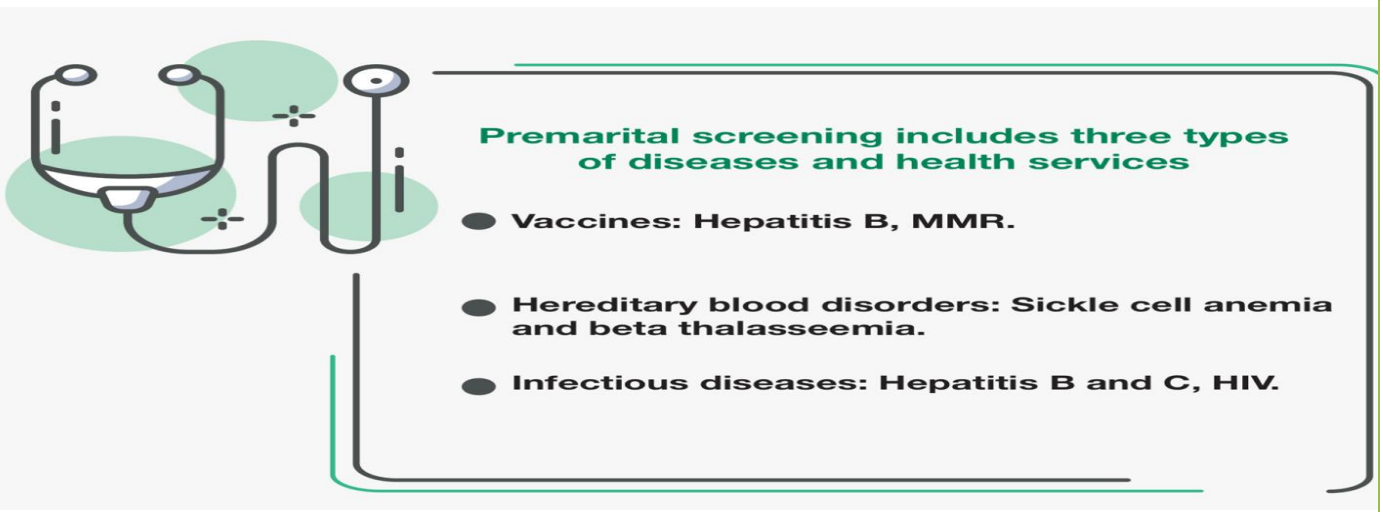
Medical consultation: An interactive process between a specialized individual (the consultation physician) who will provide medical recommendations and pertinent information in an objective manner in order to help the consultation recipient (the person planning to get married) in making the correct choices in an appropriate professional and social environment. This process aims to provide a suitable psychological atmosphere to ensure that the recipient is able to make the right decision in an atmosphere of confidence, integration, and privacy

Medical consultation clinic: The place designated for medical consultation.

Medical consultation provider: A member of the medical team specialized in medical and genetic counseling, or a person who is academically and medically trained to provide health consultation services to individuals planning to get married and families seeking information on the risks of hereditary or infectious diseases.

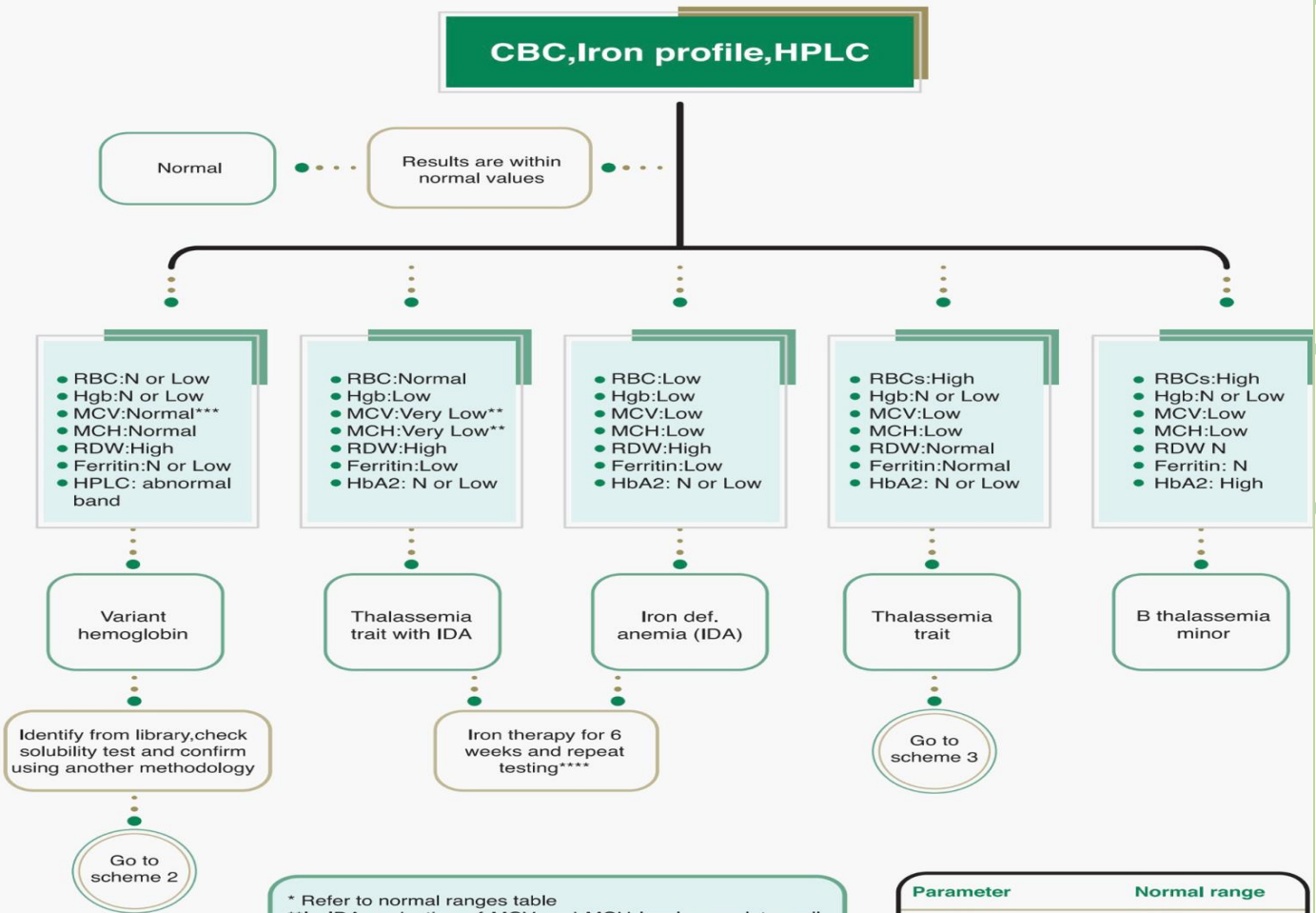
Diseases Included in premarital screening

Premarital screening is an important social and medical issue, as it prevents, Allah willing, harm to the spouse or children. Screening can detect some of the hereditary and communicable diseases, which may cause one spouse dearly because of the marriage



Assessment and Management

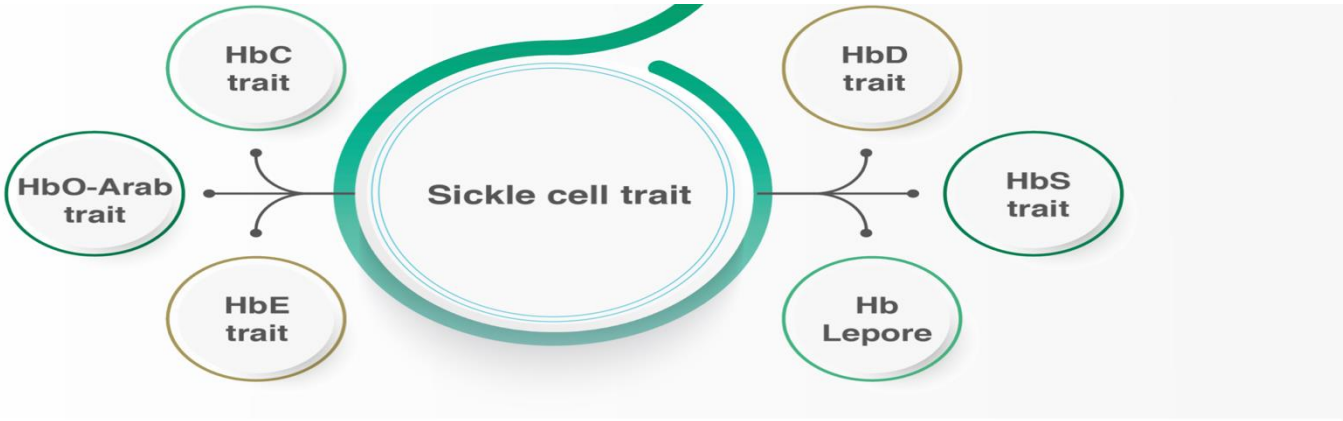
Laboratory Manual for Diagnosing Hemoglobinopathies:
• Hereditary blood disorders (sickle cell anemia and thalassemia)



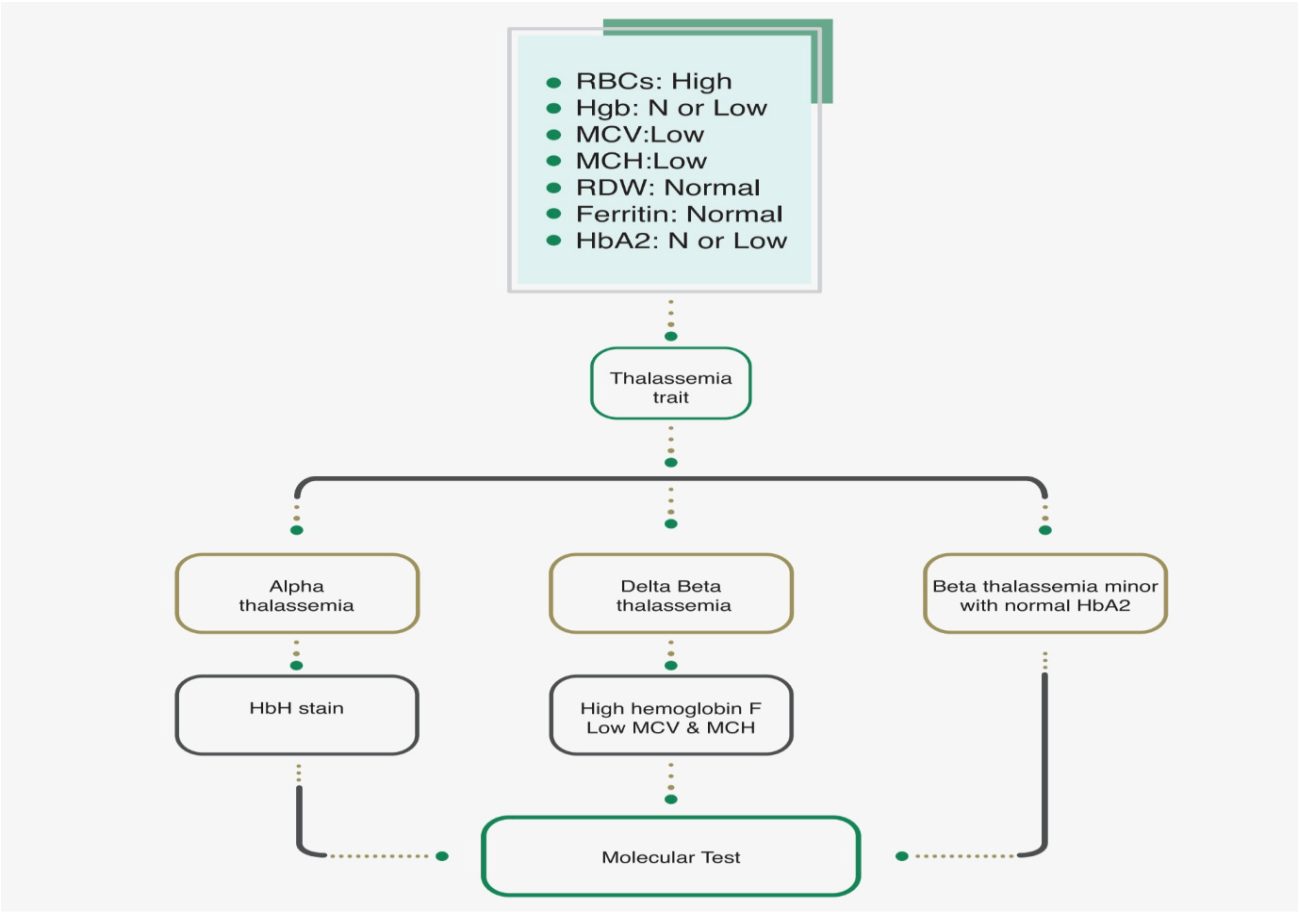
* Refer to normal ranges table
**In IDA, reduction of MCV and MCH levels correlate well with severity of anemia while in thalassemia trait, levels of MCV and MCH are disproportionally low to hemoglobin level.If IDA coexists with thalassemia trait, it leads to further reduction of MCV and MCH levels.
*** Hemoglobin E is thalassemic structural hemoglobin and usually presents with hypochromic microcytic picture.
**** Clinical decision should not be delayed if molecular test is negative or in case of latent IDA (borderline low hemoglobin level with low MCV and MCH) as this condition does not alter HbA 2 level.Moreover,mild IDA anemia (Hgb> 11g/dL and MCV> 73 fL) is less likely to lead to significant reduction of HbA 2 level.

Parameter	Normal range
RBC (10^{12} /L	4.5-5.8 3.9-5.2
Hgb (g/dL)	13 (male) 12 (female)
MCV (fL)	78-94
MCH (pg)	27-32
RDW %	11-14
HbA 2 %	<3.5
Ferritin (mcg/L)	20-400

Table: Normal ranges for RBCs indices, HbA 2 and ferritin

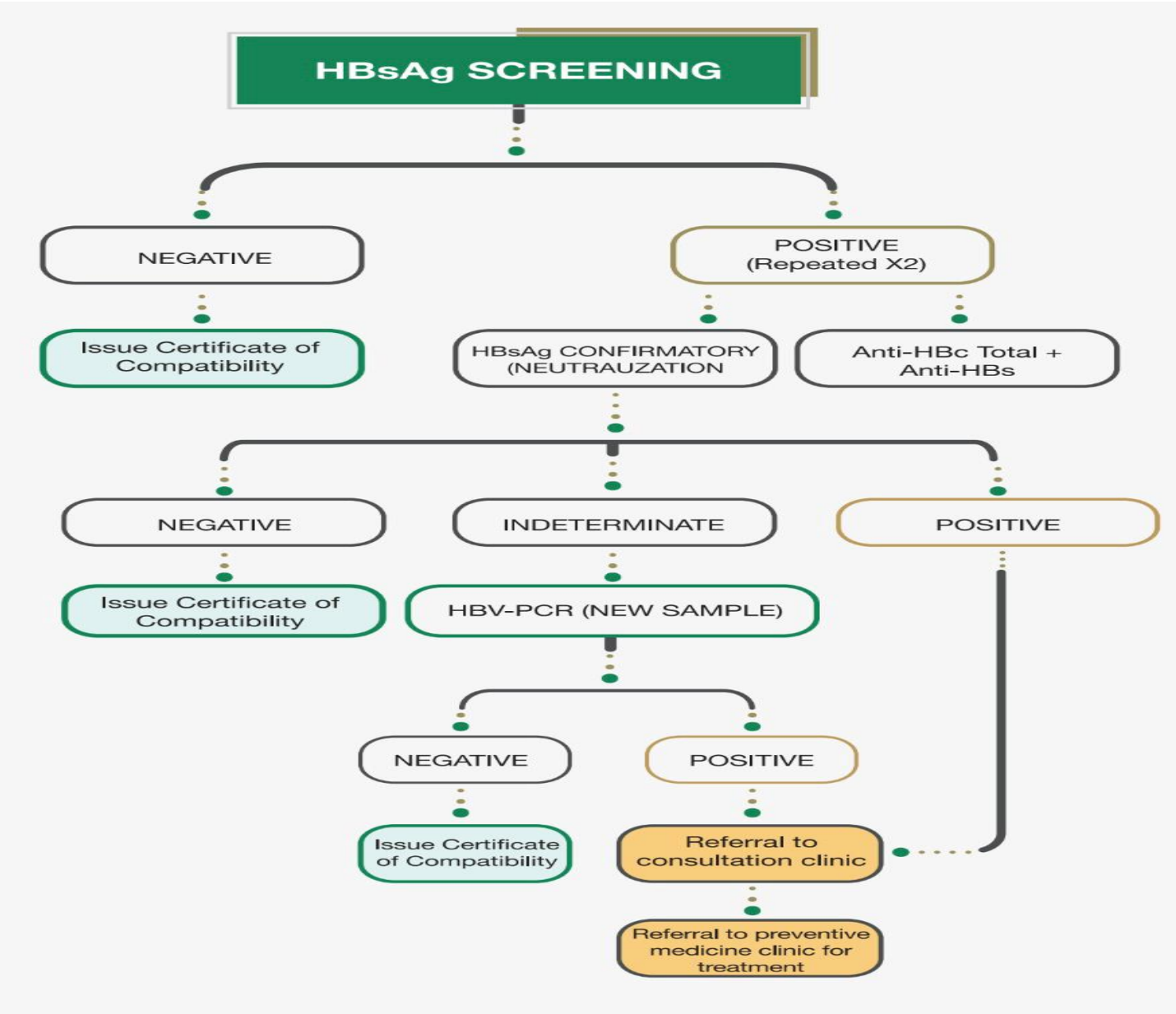


Scheme 2: Sickle Cell Anemia Overlap with Hemoglobin Variants



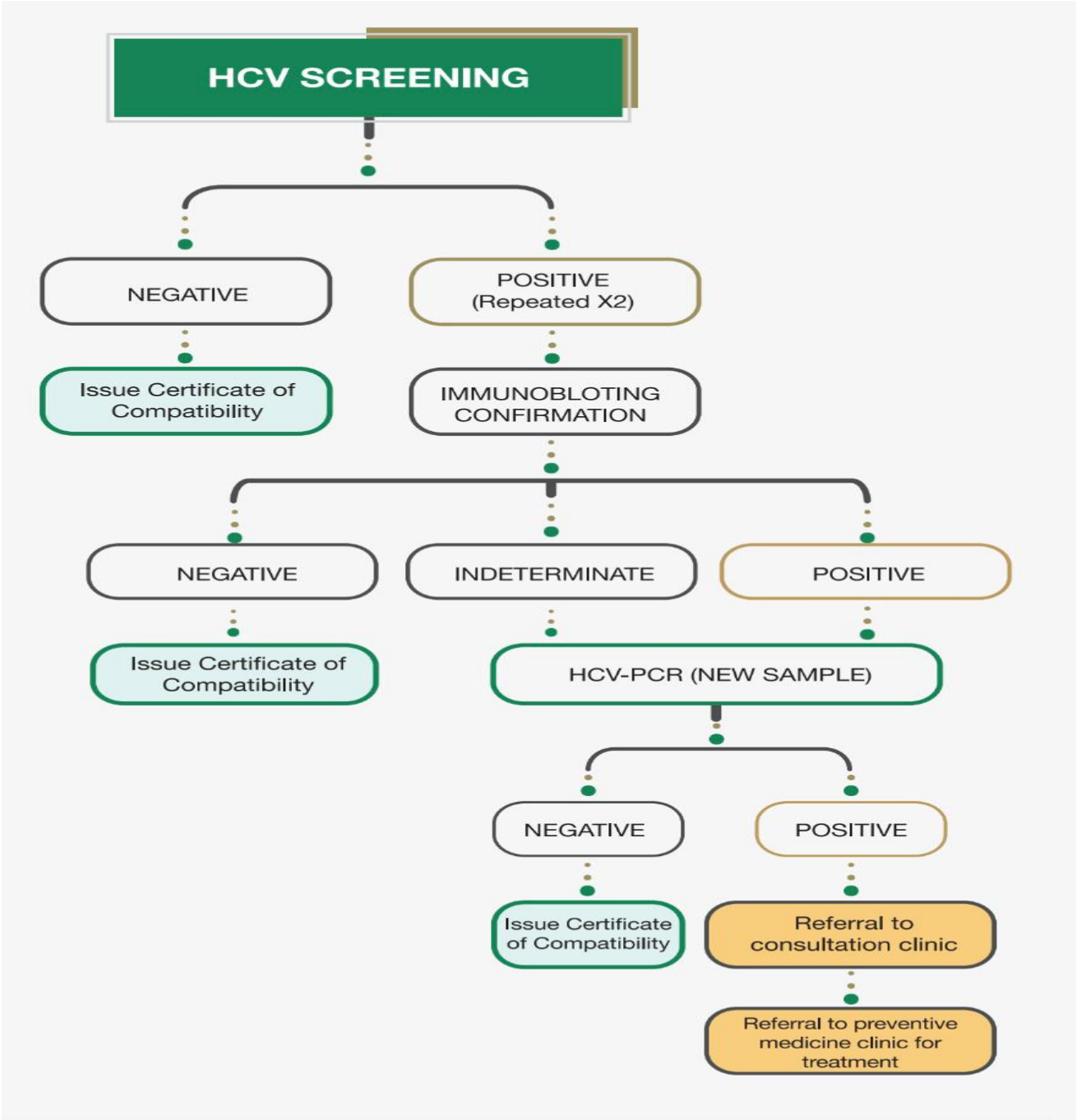
Scheme 3: Diagnosing Thalassemia Carrier State with Low or Normal Hemoglobin A2 Levels

Screening for Infectious Diseases
- Laboratory Manual for Hepatitis B Screening



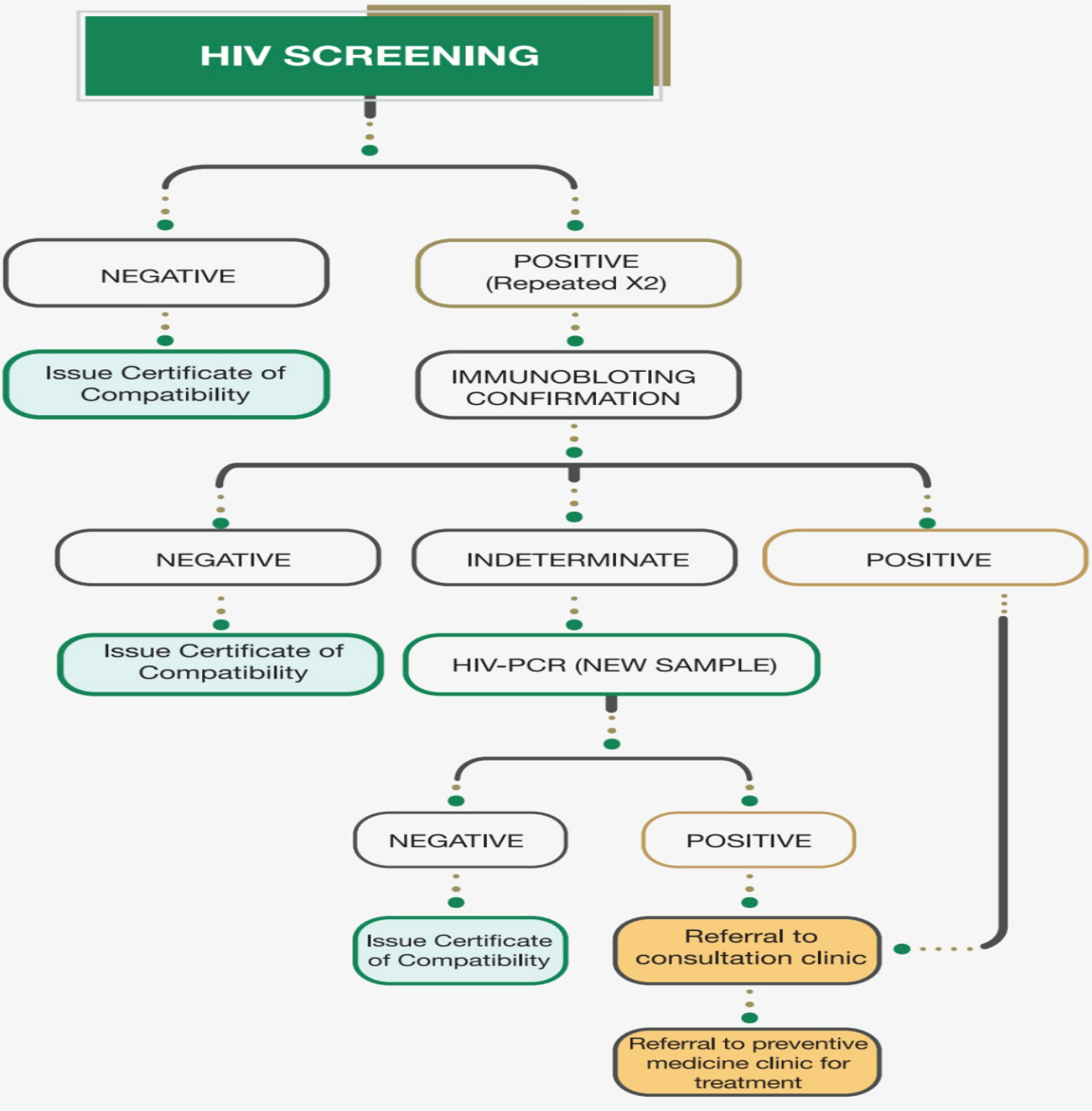
Scheme 4: Laboratory Diagnosis of Hepatitis B Virus

-Laboratory Manual for Hepatitis C Screening



Scheme 5: Laboratory Diagnosis of Hepatitis C Virus

- Laboratory Manual for HIV Screening



Scheme 6: Laboratory Diagnosis of HIV

Procedure for Administering the MMR Vaccine at Premarital Medical Evaluation Centres:

- Provide health education about the importance and benefits of vaccination.
- Confirm the absence of contraindications to administering the vaccine.
- Emphasize avoiding pregnancy for at least 28 days after receiving the vaccine.
- Signing the agreement to take the MMR vaccine and avoid pregnancy for at least 28 days after receiving it.
- If a woman planning to get married refuses the vaccine, she must sign the statement of refusal
- Enter the data of the woman planning to get married into the electronic system as per MOH instructions.
- Follow-up of premarital screening procedures.

Sickle Cell Anemia

Sickle cell anemia is a hereditary blood disorder where a defect occurs in the genes responsible for hemoglobin formation, causing red blood cells to get stuck in fine blood vessels. This diminishes the flow of blood and oxygen to the affected organ leading to appearance of the symptoms that accompany attacks in affected individuals such as severe pain, shortness of breath, and others.

Symptoms

• Recurrent attacks of pain in different parts of the body depending on the area where red blood cell sickling and blockage of fine blood vessels occurs, such as pain in the abdomen or joints or one of the limbs.
• Chronic anemia.
• Recurrent infections.
• Symptoms of malnutrition, short stature, and decreased growth velocity.
• Bone deformities.
• Malaise and fatigue.

Complications

Resulting from blockage of capillaries and early red blood cell breakdown and death, complications include:
• Thrombus formation in the heart or brain.
• Frequent infections.
• Jaundice.
• Gallstone formation.
• Visual disturbance and loss of vision.
• Delayed growth in children.

Treatment

• The main aim of treatment in sickle cell anemia is to lessen the frequency of acute crisis episodes in the affected person in order to limit the development of complications, reduce pain, and improve the person's ability to live with the disease.
• People with sickle cell anemia need continuous care in order to lessen the frequency of complications and health deterioration.
• People with sickle cell anemia are given folic acid pills to help red blood cell production.
• During crisis episodes, pain is managed with analgesic medications and hydrating the patient's body.
• For pain management, some cases respond to over-the-counter analgesic medications, while others require stronger pain medications like morphine and meperidine and similar drugs administered in the hospital under medical supervision.
• Hydroxyurea is used in some cases as a preventive measure to prevent crises from happening, especially symptoms related to the respiratory system such as chest pain and shortness of breath.
• Routine vaccinations are a must, especially for children with the disease, in addition to annual seasonal vaccines such as influenza vaccine, to prevent infection.
• Patients may need to receive blood transfusions regularly and during acute crises.
• Eyes may be affected in persons with sickle cell anemia, and complications may occur leading to loss of vision; and so, regular follow-up and examination by an ophthalmologist is important.
• Bone marrow transplant can be considered for treatment in persons with sickle cell anemia

Thalassemia

Thalassemia, or Mediterranean anemia, is a hereditary blood disorder that leads to diminished production of hemoglobin and red blood cell destruction. It is known as Mediterranean anemia because of its high prevalence in that region, and it results from a defect in the genetic makeup of hemoglobin.

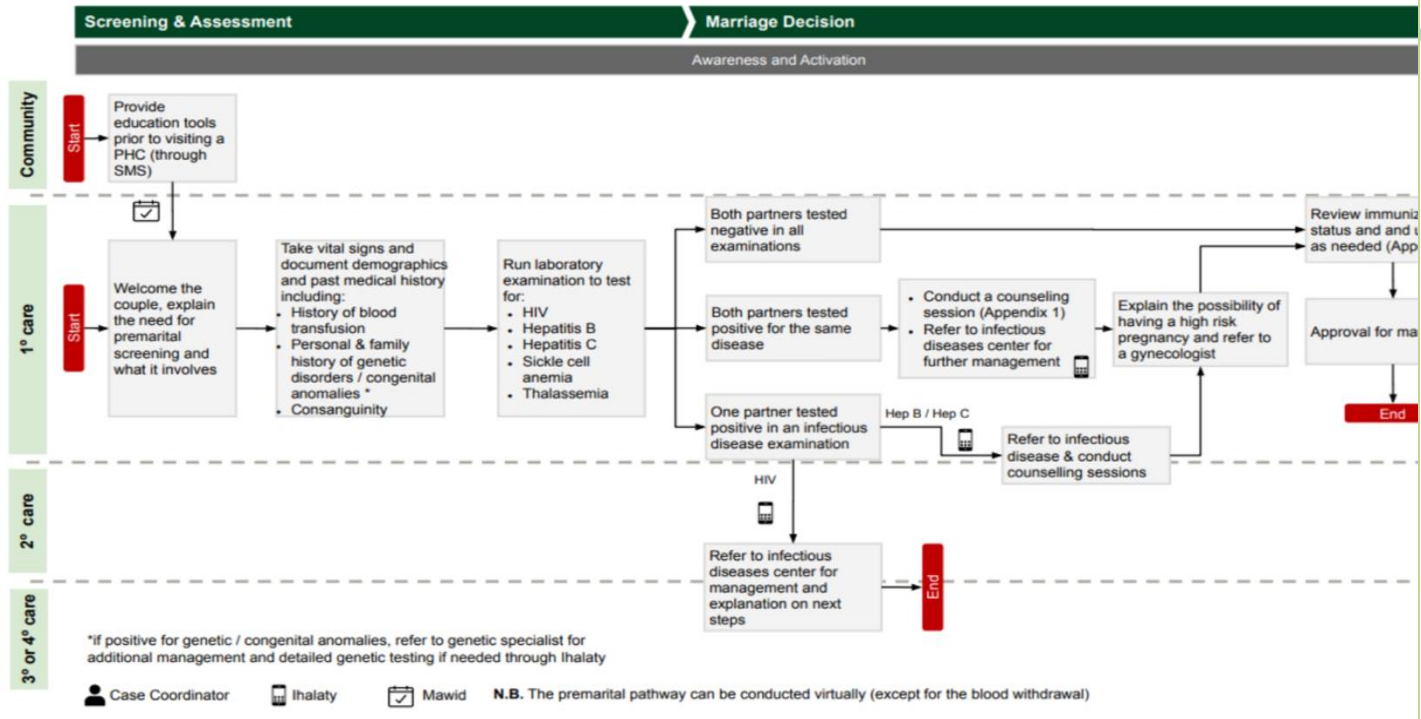
Thalassemia Types and Symptoms

The hemoglobin particle is made up of four protein chains: two alpha and two beta chains. In alpha thalassemia, fewer alpha chains are produced, and in beta thalassemia, fewer beta chains are produced.

Treatment

• Patients with thalassemia major and hemoglobin H disease need continuous regular care to prevent the many complications of thalassemia such as recurrent infections and health deterioration, which can lead to death if not appropriately managed.
• Affected persons require blood transfusion on a regular basis to raise the number of red blood cells in the blood and avoid anemia.
• Folic acid tablets are given to affected persons to help red blood cell production.
• Some persons with thalassemia may need to get rid of excess accumulated iron resulting from the continuous breakdown of red cells, which can be accomplished with the medication Desferal.
• Some persons with thalassemia major require splenectomy.
• Bone marrow transplant can be considered for patients who are diagnosed early before complications develop.

R3 Premarital Screening Pathway:



APPROVL			
	Name:	**Position:**	**Signature:**
Prepared By:	Dr. Salwa Al Malki	FM Senior Registrar	
Reviewed and Approved By:	Dr. Mansoor Allajhar Dr. Musa Althwayee Dr. Ahmed Al Zahrani Dr. Hajar Al Suma Dr. Ahlam Al Harbi	FM Consultants	

Adopted from;

Saudi MOH Guidelines