Part II
MANAGEMENT OF THALASSEMIA

1. Initial Management of Newly Diagnosed Infant

Principles

- To promptly establish the correct diagnosis for the infant with thalassemia.
- To promptly start an appropriate treatment program for the infant with thalassemia.
- To provide education and psychosocial support tailored to the education level, culture and language of the family.

Recommendation

- Affected newborns/infants should be referred promptly to a comprehensive thalassemia program and/or to a pediatric hematologist.
- Treatment should be tailored to the clinical phenotype.
- Qualified experienced professionals should discuss with the family the diagnosis, management and overall psychosocial impact on the child and family in an open, sensitive, and culturally appropriate manner.
- A written summary of the diagnosis, the treatment plan, and the discussions held should be documented and distributed to the family practitioner or pediatrician and the family.

Background

Transfusion-dependent and non-transfusion dependent thalassemias are lifelong medical conditions that require continuous care by a qualified experienced team of health professionals. Trait carriers are usually asymptomatic and they do not require any specific medical care but they should be aware of their status for reproduction purposes.
Early diagnosis of transfusion dependent thalassemia in neonates and children ensures prompt monitoring and treatment for patients who require chronic transfusion support for normal growth and development.

Interventions

Investigation and Diagnosis

- Diagnosis of a child with a thalassemia disorder should be done as early as possible after birth.
- Neonatal screening programs, where present, should be able to identify affected infants and refer them promptly to specialist centre.
- Where available, the diagnosis of a serious thalassemia syndrome should be predicted from antenatal screening of parents, and may be established by prenatal diagnosis or by neonatal testing.
- Initial diagnostic investigations should include:
    - A complete blood count and a blood smear.
    - Hemoglobin analysis by electrophoresis or high performance liquid chromatography (HPLC).
    - Molecular genetic analysis for both alpha and beta globin gene.
    - Extended red blood cell phenotype if transfusions are anticipated. Red blood cell genotyping should be considered if available.
    - Parents and siblings should be tested if no prior testing has been performed.
- Affected newborns/infants should be referred promptly to a comprehensive thalassemia program and/or to a

pediatric hematologist.
- In symptomatic, previously unidentified infants with suspected thalassemia, investigations and initial assessment should be done immediately at the specialist centre.

Treatment

- The child's clinical course should be closely monitored since the clinical phenotype cannot always be accurately predicted from the genotype. Co-inheritance of alpha-thalassemia mutations and foetal hemoglobi level can modify the clinical phenotype.
- Treatment should be tailored to the clinical phenotype. Figure 1 summarizes the Clinical and laboratory aspects to consider appropriateness for initiating a transfusion program and management of infants with new Thalassemia diagnosis.

Education

- Qualified experienced professionals should discuss with the family the diagnosis, management and overall psychosocial impact on the child and family in an open, sensitive, and culturally appropriate manner.
- Both parents should be given verbal and written information about the diagnosis and management, and given the opportunity to ask questions. An interpreter may be necessary to communicate in the family's language of first choice, if English or French is not the primary language.
- The key contact specialist nurse should meet and exchange contact information with the family.
- The family should be given information on regional support groups, if available.
- A written summary of the diagnosis, the treatment plan, and the discussions held should be documented and distributed to the family practitioner or pediatrician and the family.

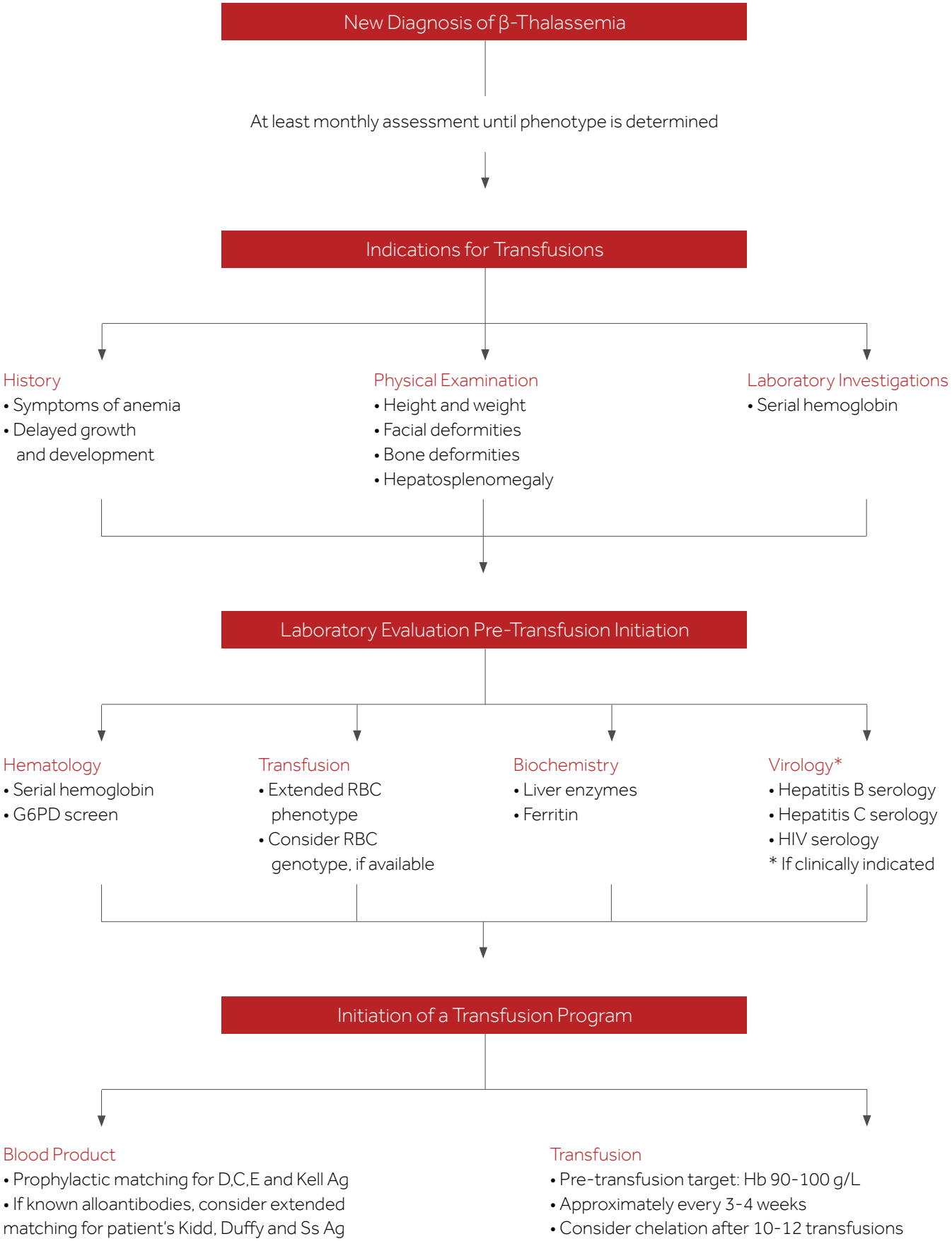

Figure 1: Clinical and laboratory aspects to consider appropriateness for initiating a transfusion program