Mother of a one-year-old child diagnosed to have beta thalassemia major is waiting to talk to you

Your patient is an actor but you are NOT. Do what you could do as a third year student. Try to gather knowledge in advance.

You have not met this mother before. She is waiting to talk to you regarding the results of the HPLC report. This one-year-old child has been investigated for anaemia since 8 months of age. All the previous blood tests have suggested that the child is likely to have beta thalassemia major.
Her nephew (sisters daughter) died at the age of 2 years with similar illness.

Facts about HPLC – HPLC can be considered as the conclusive investigation in the diagnosis of thalassemia.

Facts about beta thalassemia – Thalassemia are a group of diseases manifest as chronic haemolytic anaemia due to lack or inadequate production of either alpha or beta globin chains. In this case the defects is in the production of beta chain; hence it is called beta thalassemia major. The extent of the deficiency of production may vary from total absence to a slight reduction in the production. This case has the severe deficiency resulting in MAJOR thalassemia.

The disease thalassemia is inherited as an autosomal recessive disease. That means that both parents contribution of defective genes at the time of conception is essential for the birth of a baby with thalassemia major. This implies that in a case of a thalassemia major both parents are carriers of the disease. In this case both parents are carriers. There is a 25% risk of having a similarly affected child at the next pregnancy also.

This child with thalassemia major will need regular life long blood transfusion at intervals varying from 3 weeks to 6 weeks. After giving about 10 blood transfusions he is likely to get iron over load as a result of iron in the transfused blood.

Iron in excess is toxic to human body. Usual mechanisms of keeping iron non-toxic by binding to a transporting protein get exhausted resulting in the appearance of free iron in blood. These free iron molecules release electron-producing free radicals that damage internal organs. Worst affected are the heart, liver and all the endocrine organs. The iron over load can be managed by giving regular life long chelation regimen. The ministry of health provides theses very expensive drugs free of any charge. Regular blood transfusion and chelation gives the hope of a normal life to patients with thalassemia major.