How to get the most from...

THE COOLEY'S ANEMIA FOUNDATION'S
2017 PATIENT-FAMILY CONFERENCE

Dr Richard Ward,
Adult Hematologist, Toronto

“You need to change your dressing.”

OR

“We need a stool sample from you.”

OR

patients with low

HEALTH LITERACY...

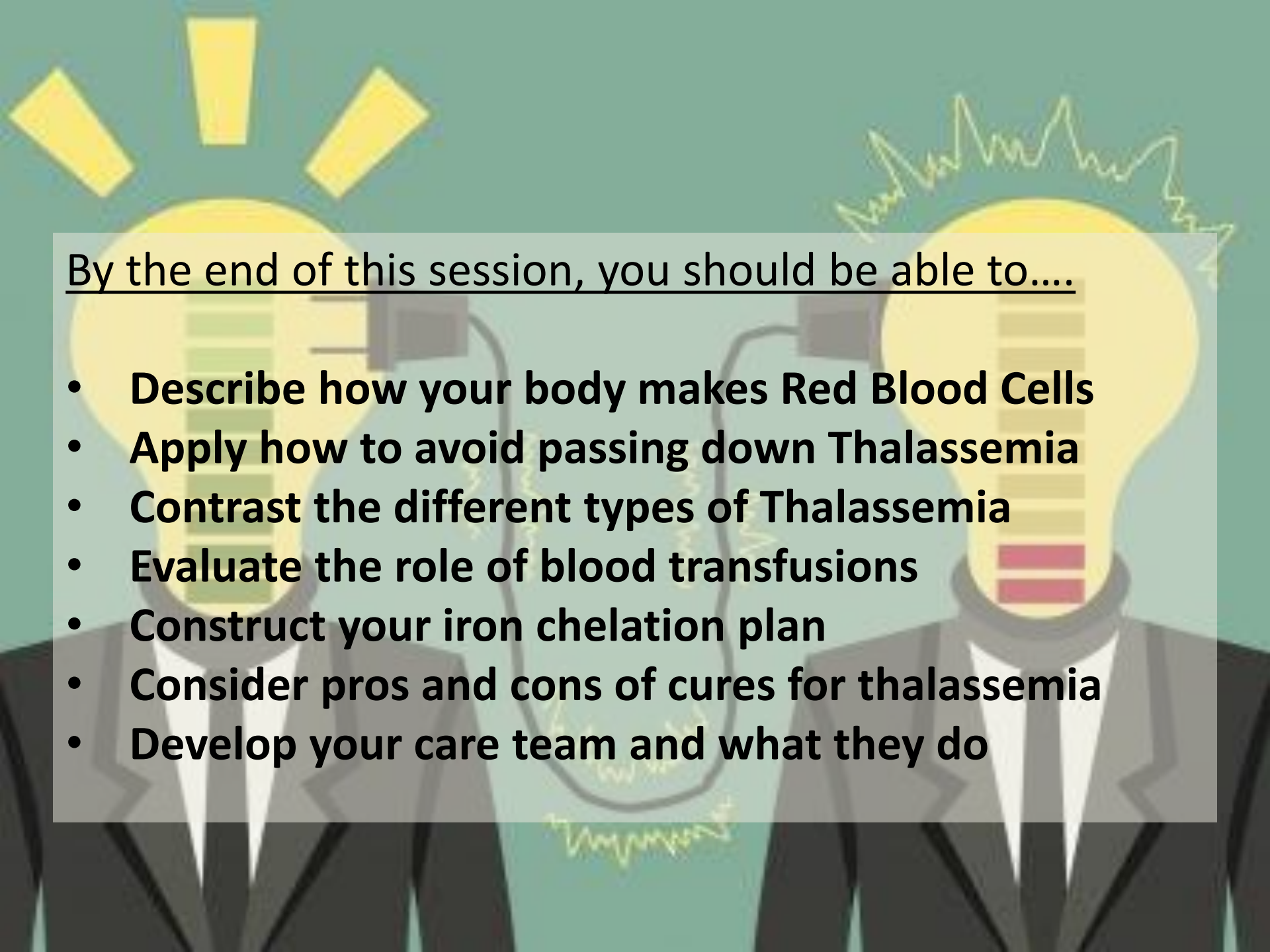
Are more likely to visit an **EMERGENCY ROOM**

Have more **HOSPITAL STAYS**

Are less likely to follow **TREATMENT PLANS**

Have higher **MORTALITY RATES**

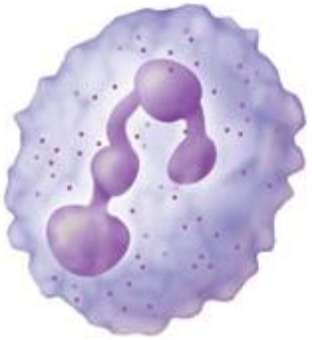
www.cdc.gov/phpr



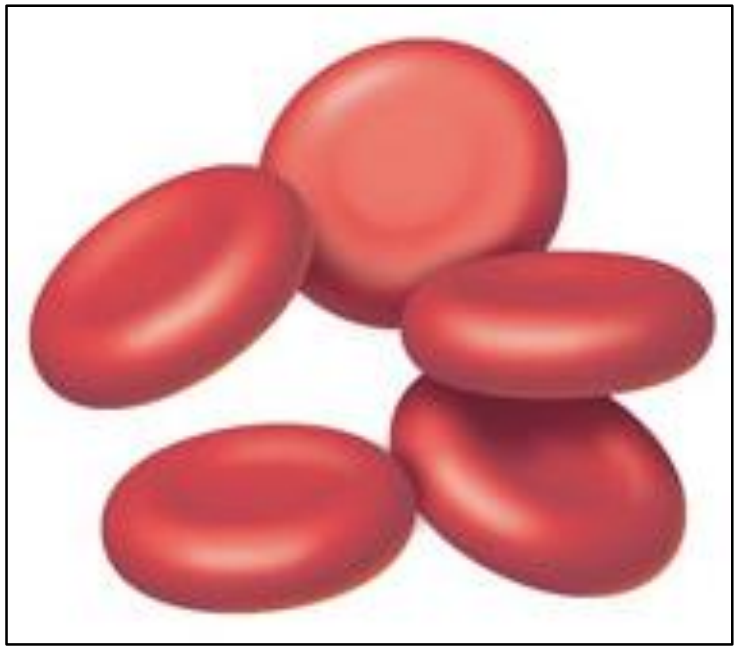
By the end of this session, you should be able to….

- **Describe how your body makes Red Blood Cells**
- **Apply how to avoid passing down Thalassemia**
- **Contrast the different types of Thalassemia**
- **Evaluate the role of blood transfusions**
- **Construct your iron chelation plan**
- **Consider pros and cons of cures for thalassemia**
- **Develop your care team and what they do**

Blood Stem Cell

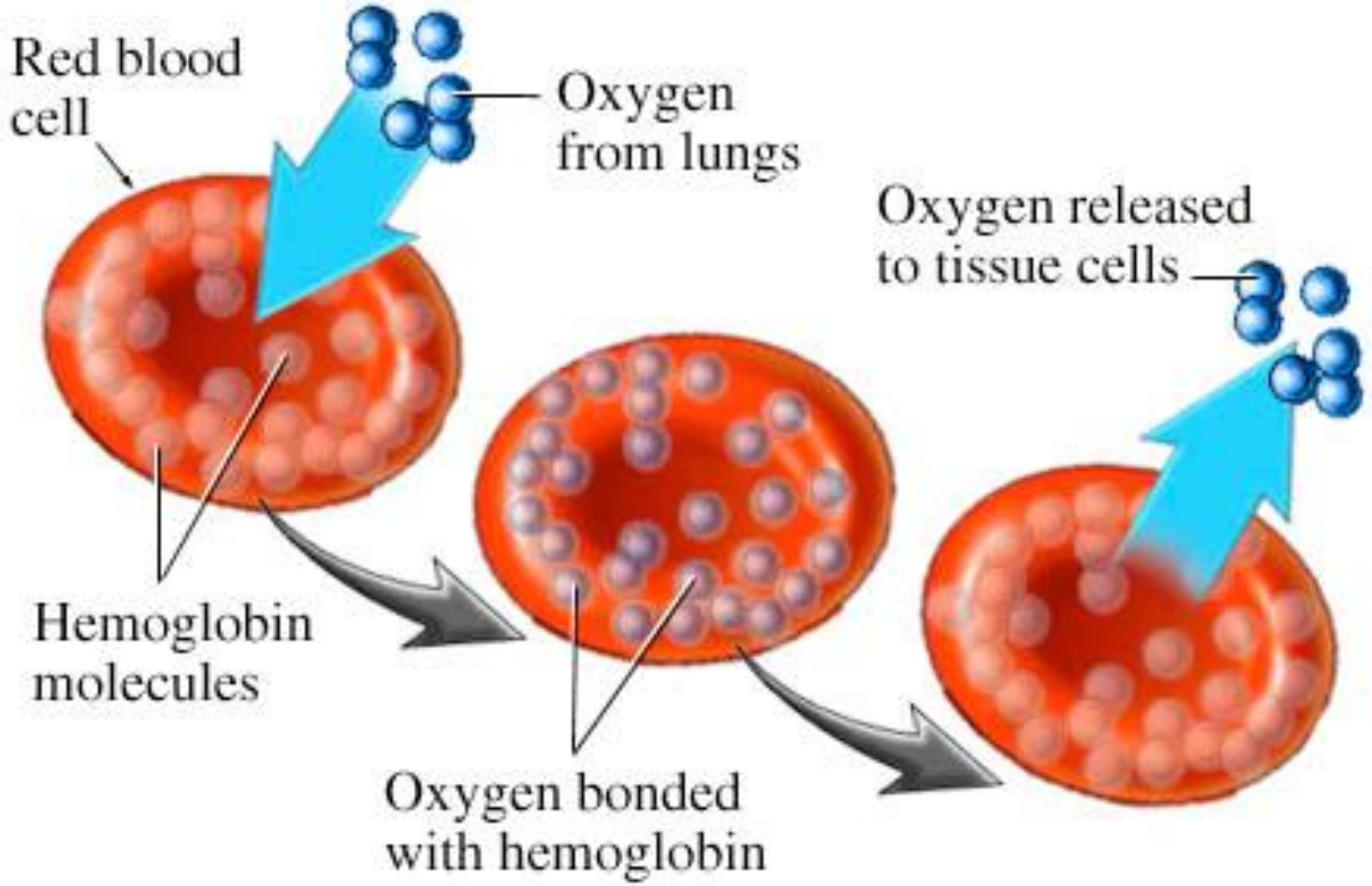


White Blood Cells (WBC)
fight infection



Red Blood Cells (RBC)
contain Hemoglobin (Hb)

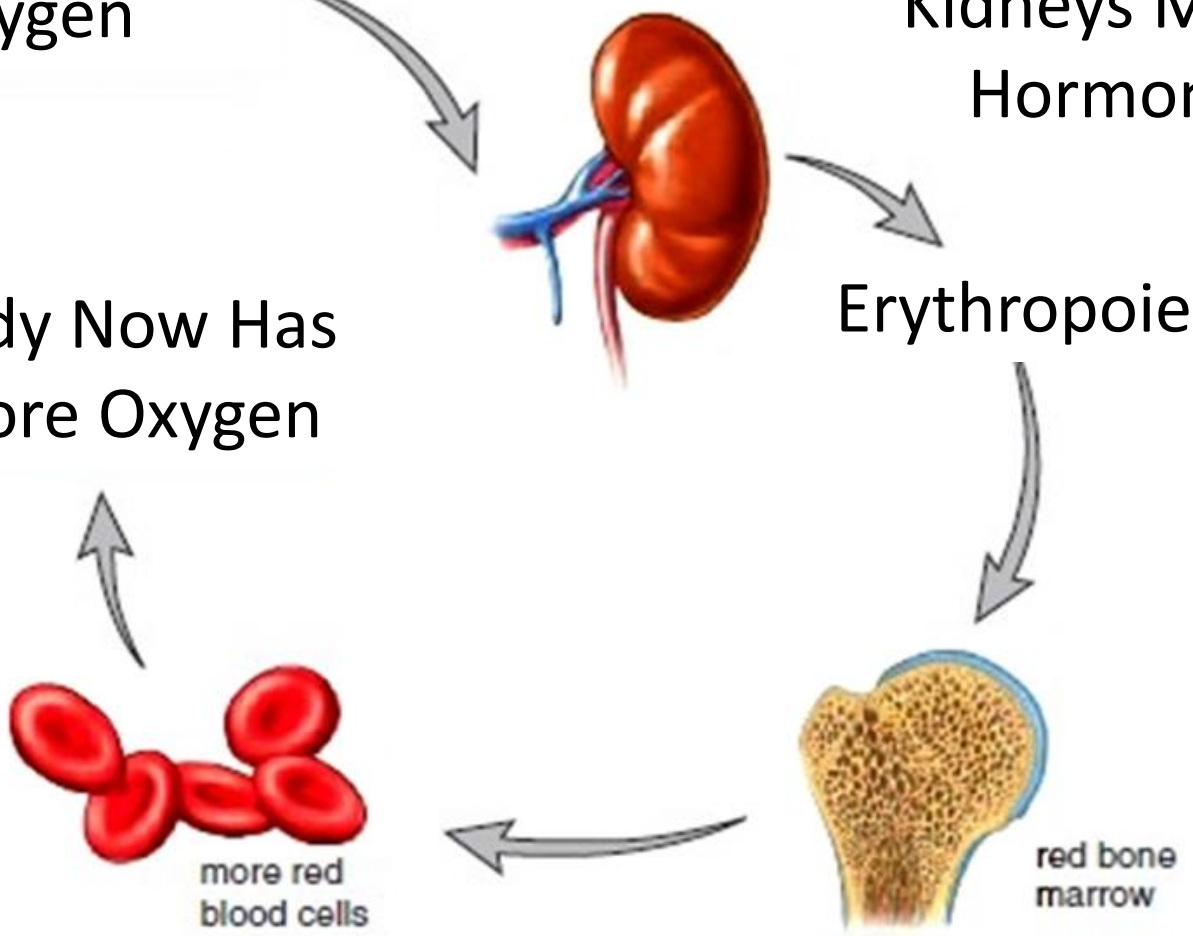
Platelets
stop bleeding


Red blood cell
Oxygen from lungs
Oxygen released to tissue cells
Hemoglobin molecules
Oxygen bonded with hemoglobin

Body is Low in Oxygen

Kidneys Make Hormone

Erythropoietin (Epo)

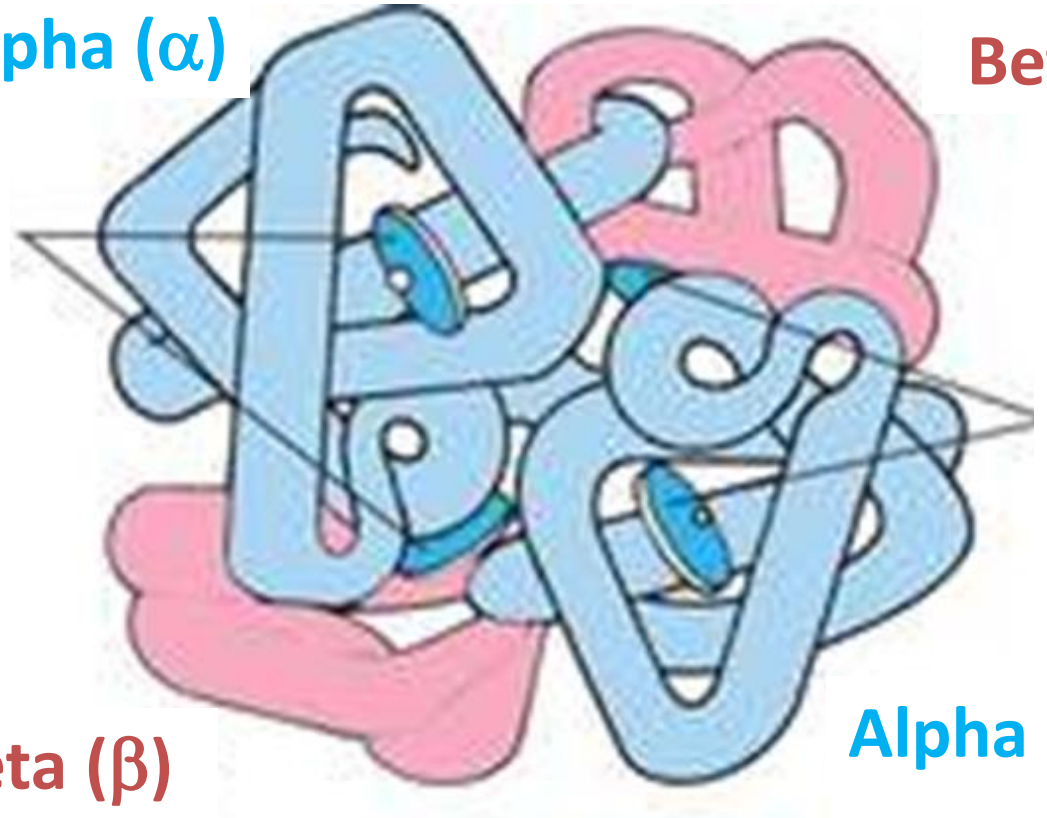


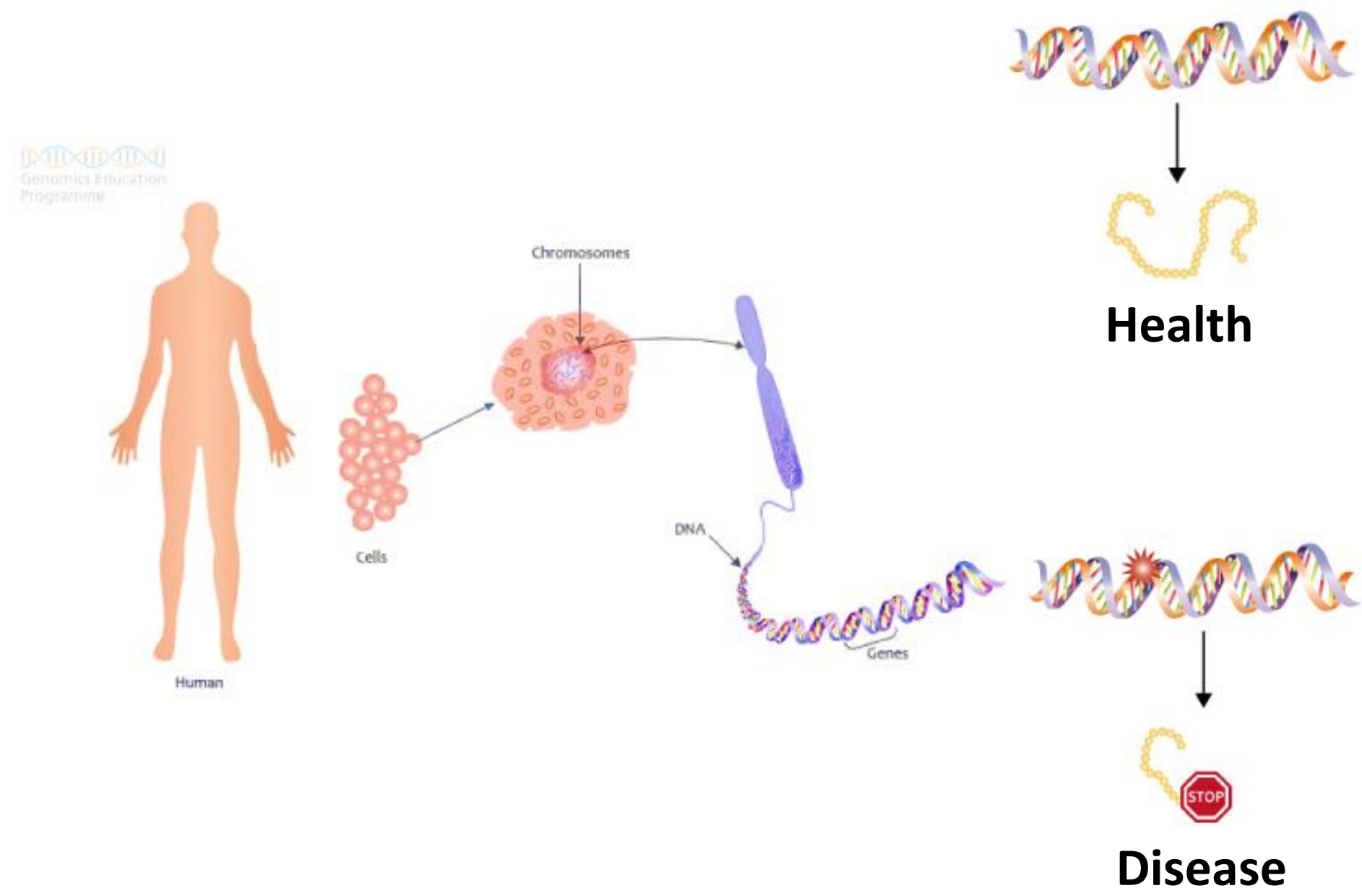
Bone Marrow Makes More RBCs

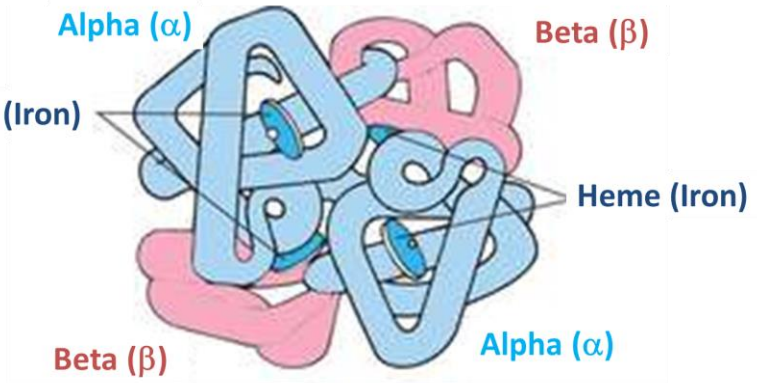
Body Now Has More Oxygen

LOOK INSIDE!

Hemoglobin




Genomics Education Programme
Chromosomes
Cells
DNA
Genes
Human
Health
STOP
Disease

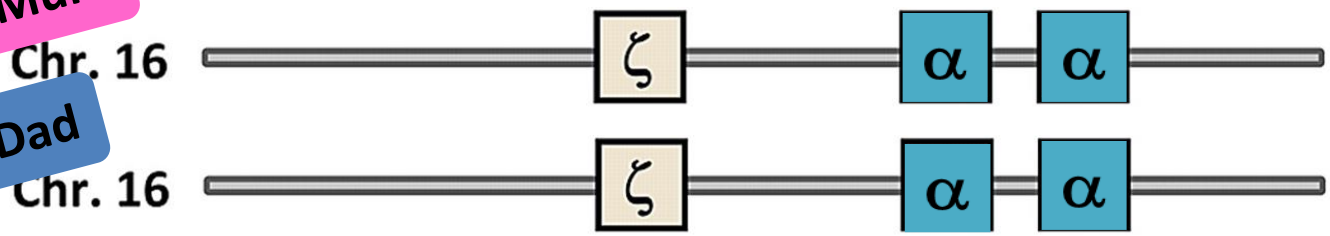


LOOK INSIDE!

Hemoglobin

from Mum

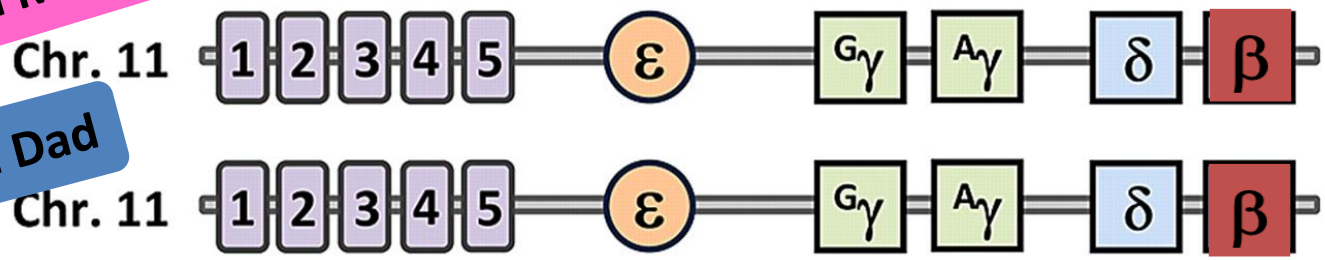
from Dad



= 4 alpha genes code for the alpha chains

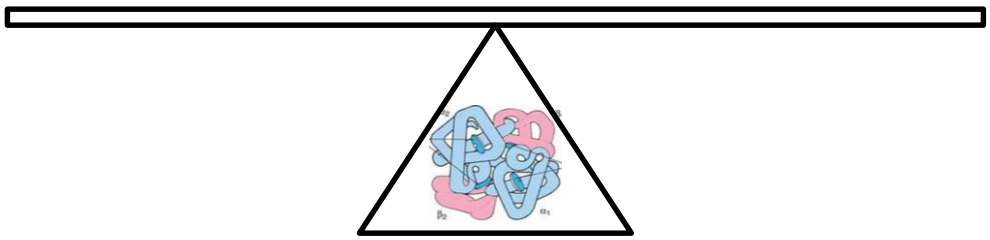
from Mum

from Dad



= 2 beta genes code for the beta chains

α = normal = β



β

α

= beta thalassemia

(less β chains, too many α chains)

α

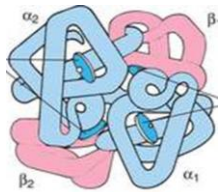
β

= alpha thalassemia

(less α chains, too many β chains)

Anemia……

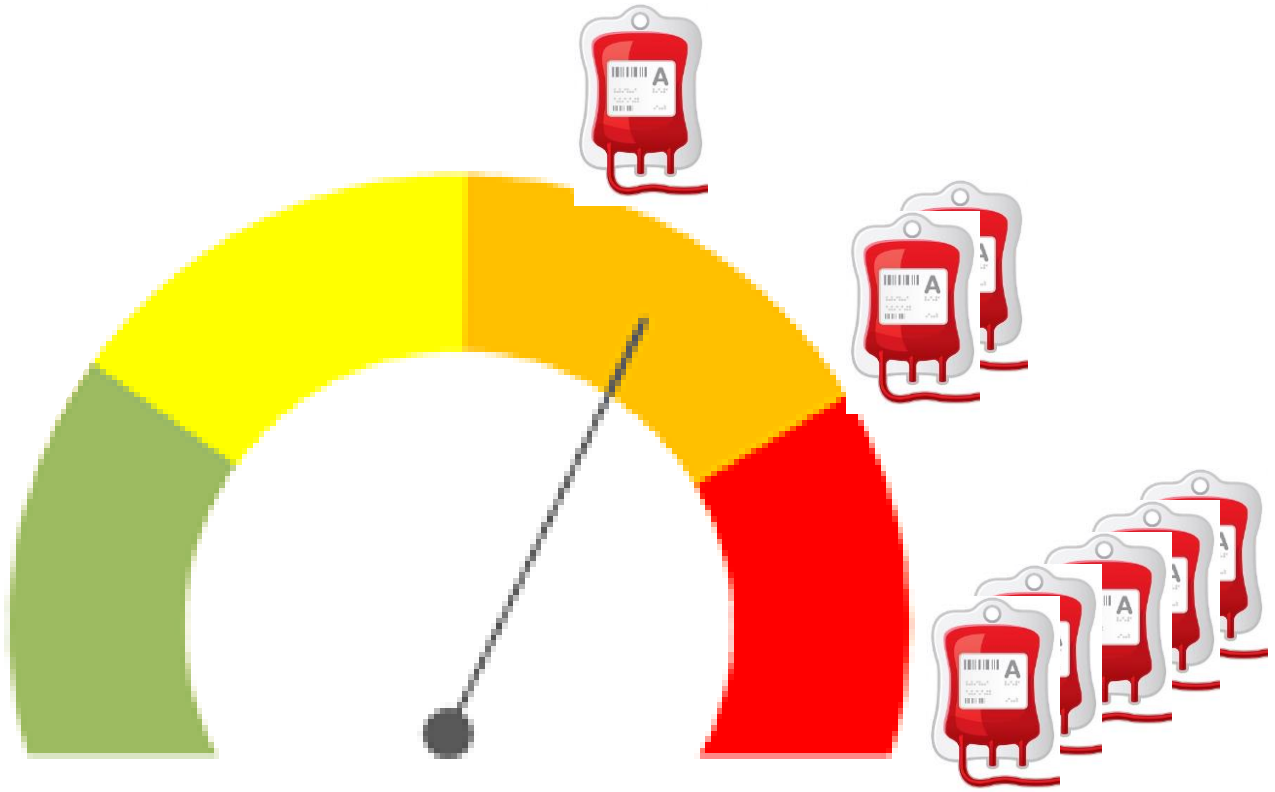
Fewer: 

Less: 

♀ **<120** g/L

♂ **<140** g/L

…so, less Oxygen carried around the body



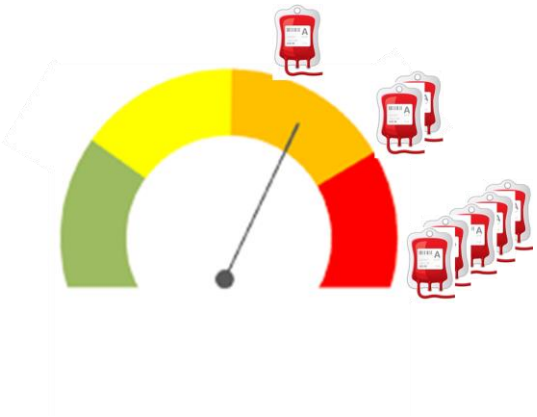
No Thalassemia

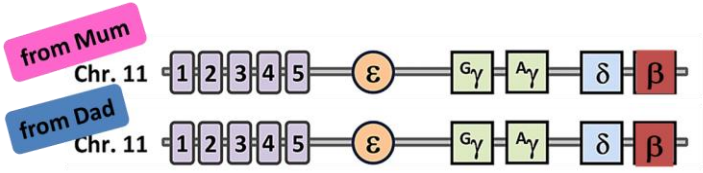
Thalassemia Trait

Thalassemia Intermedia or
Non-Transfusion-Dependent-Thalassemia

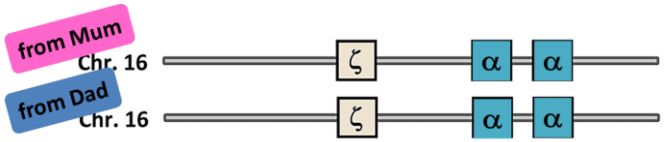
Thalassemia Major

Thalassemia Trait





- not making as many beta chains from **<u>one</u>** of the 2 genes
- the other beta gene is working fine

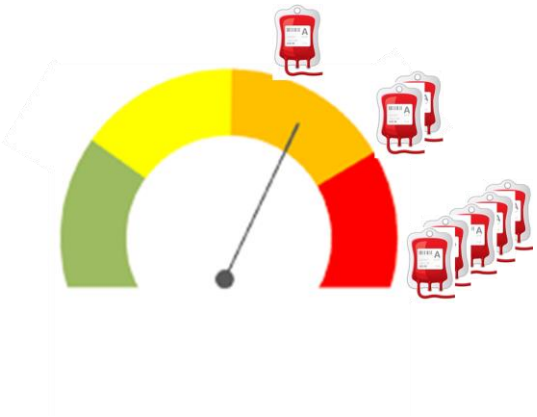


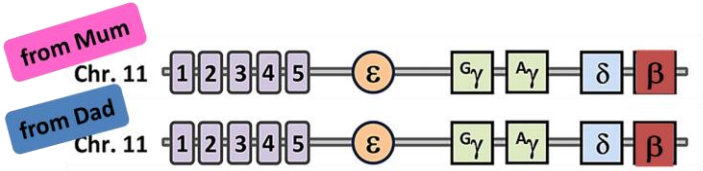
- not making as many alpha chains from **<u>one or two</u>** of the 4 genes
- the other two or three genes are working fine

1. Not a disease

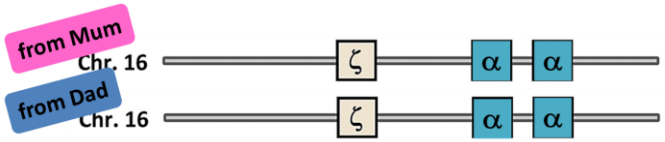
2. Genetic counseling

Thalassemia Intermedia or Non-Transfusion-Dependent-Thalassemia





- not making as many beta chains from **one or both** of the 2 genes
- the beta genes are still making **some** beta chains

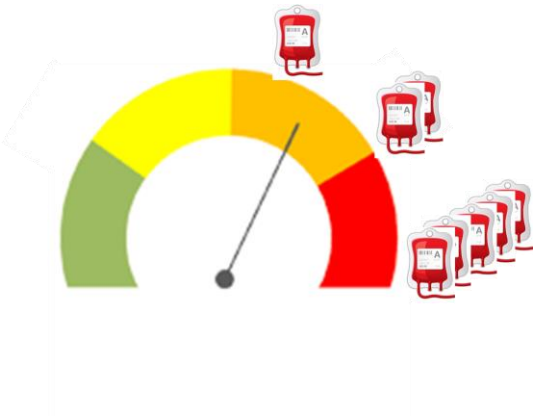


- not making as many alpha chains from **three** of the 4 genes
- the other gene is still making **some** alpha chains
- called **HbH Disease** as makes some Hb made up of β x4

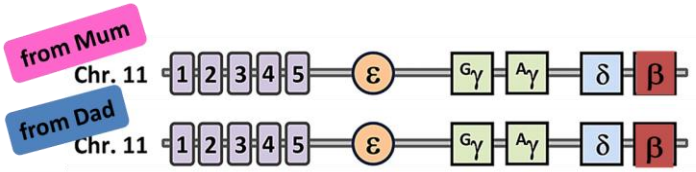
1. TI/NTDT causes problems

2. Genetic counseling

Thalassemia Intermedia or
Non-Transfusion-Dependent-Thalassemia

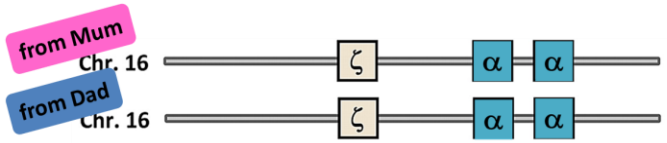


HbE



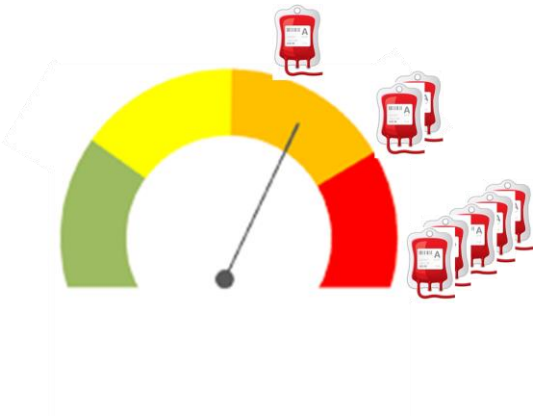
- a different form of beta chain is made
- it does not last long, so not much of it in the body
- causes Thalassemia

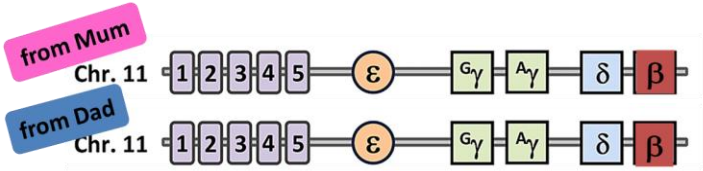
Hb Constant Spring



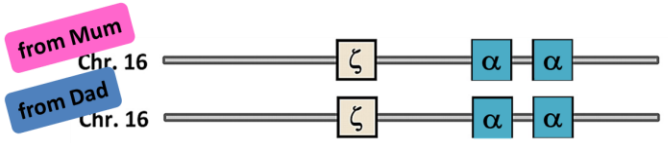
- a **different form** of alpha gene change (mutation)
- **more severe** than regular alpha thalassemia gene changes

Thalassemia Major





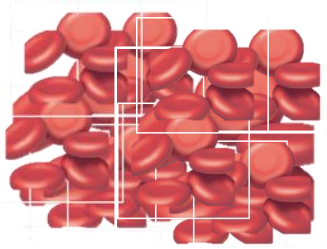
- not making beta chains from **both** of the 2 genes



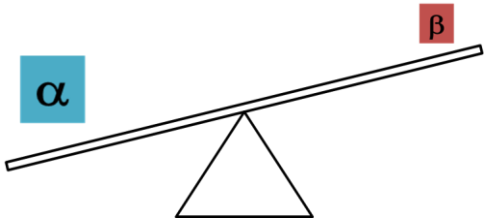
- not making alpha chains from **all four** genes
- called **HbBarts (Barts Hydrops Fetalis Syndrome)** as makes some Hb made up of γ x4

1. TM causes problems

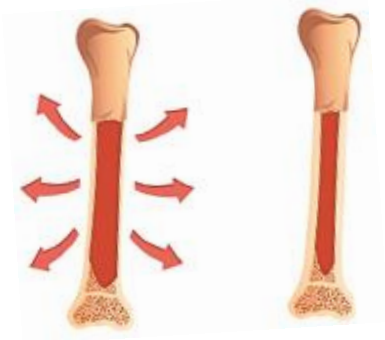
2. Genetic counseling

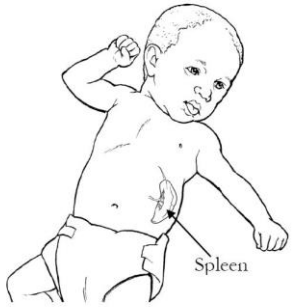


βTHAL.



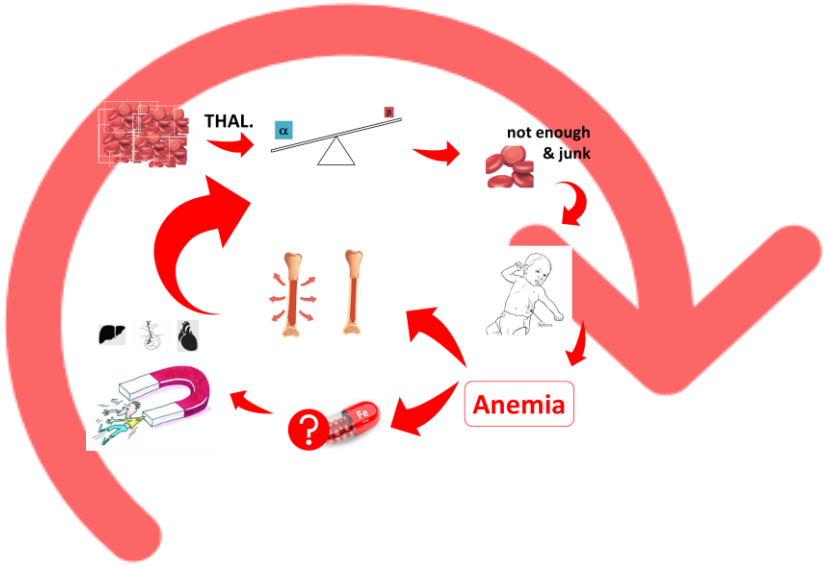
not enough & junk





Anemia

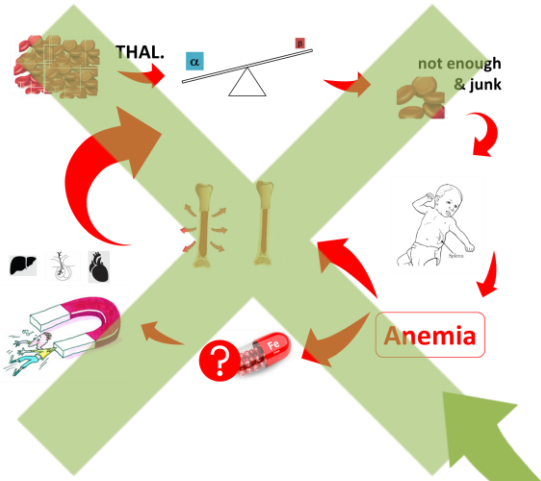
Thalassemia Intermedia or Non-Transfusion-Dependent-Thalassemia



Do not need blood each month

- **Weak bones (osteoporosis)**
- **Large bones**
- **Lumps of blood tissue grow**
- **Large spleen and liver**
- **Too much iron in the liver**
- **Blood clots**
- **High blood pressure in the lungs**

Thalassemia Major



Need lots of blood each month

- **Bones healthier**
- **Smaller lumps of blood tissue**
- **spleen and liver not big**
- **Low risk of blood clots and lung pressure**

When Do I Start to Get Blood?

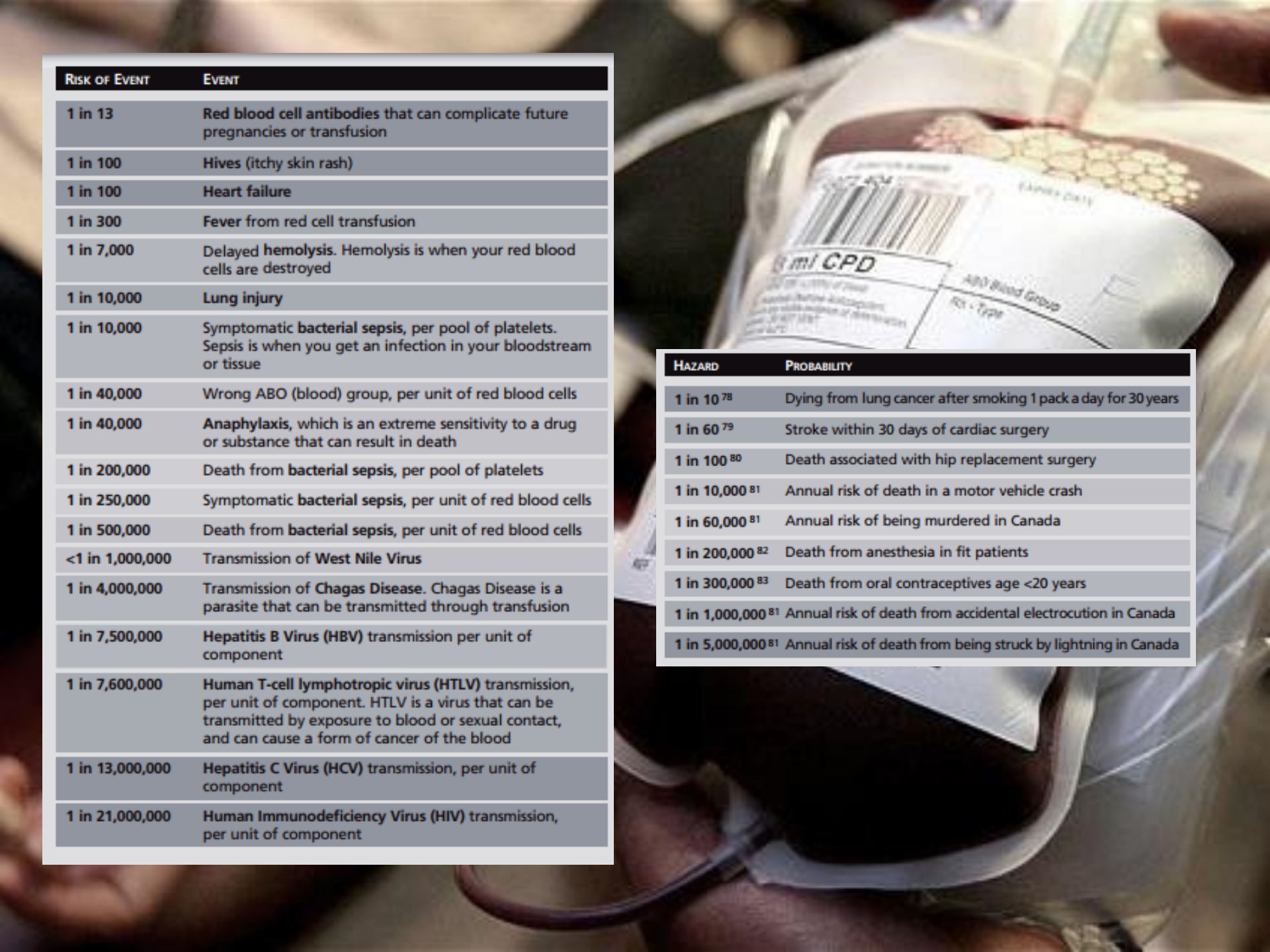
- at a few months of age if thalassemia major
- if problems with anemia such as:
  - low energy, not growing, large spleen or bones

How Often Will I Come For Transfusion?

- every 2-5 weeks
- some patients with NTDT choose to only have when needed (not optimal)

How Will I Get Transfusions?

- crossmatch test 1-3 days ahead of date
- PSL (normal), PVR (plasma volume reduced), or washed units
- volume by weight or whole units to keep Hb>100



Risk of Event	Event
1 in 13	**Red blood cell antibodies** that can complicate future pregnancies or transfusion
1 in 100	**Hives** (itchy skin rash)
1 in 100	**Heart failure**
1 in 300	**Fever** from red cell transfusion
1 in 7,000	Delayed **hemolysis**. Hemolysis is when your red blood cells are destroyed
1 in 10,000	**Lung injury**
1 in 10,000	Symptomatic **bacterial sepsis**, per pool of platelets. Sepsis is when you get an infection in your bloodstream or tissue
1 in 40,000	Wrong ABO (blood) group, per unit of red blood cells
1 in 40,000	**Anaphylaxis**, which is an extreme sensitivity to a drug or substance that can result in death
1 in 200,000	Death from **bacterial sepsis**, per pool of platelets
1 in 250,000	Symptomatic **bacterial sepsis**, per unit of red blood cells
1 in 500,000	Death from **bacterial sepsis**, per unit of red blood cells
<1 in 1,000,000	Transmission of **West Nile Virus**
1 in 4,000,000	Transmission of **Chagas Disease**. Chagas Disease is a parasite that can be transmitted through transfusion
1 in 7,500,000	**Hepatitis B Virus (HBV)** transmission per unit of component
1 in 7,600,000	**Human T-cell lymphotropic virus (HTLV)** transmission, per unit of component. HTLV is a virus that can be transmitted by exposure to blood or sexual contact, and can cause a form of cancer of the blood
1 in 13,000,000	**Hepatitis C Virus (HCV)** transmission, per unit of component
1 in 21,000,000	**Human Immunodeficiency Virus (HIV)** transmission, per unit of component

Hazard	Probability
1 in 10 [78]	Dying from lung cancer after smoking 1 pack a day for 30 years
1 in 60 [79]	Stroke within 30 days of cardiac surgery
1 in 100 [80]	Death associated with hip replacement surgery
1 in 10,000 [81]	Annual risk of death in a motor vehicle crash
1 in 60,000 [81]	Annual risk of being murdered in Canada
1 in 200,000 [82]	Death from anesthesia in fit patients
1 in 300,000 [83]	Death from oral contraceptives age <20 years
1 in 1,000,000 [81]	Annual risk of death from accidental electrocution in Canada
1 in 5,000,000 [81]	Annual risk of death from being struck by lightning in Canada

Take iron seriously for a healthy

Adherence is…..

- Lifestyle
- Commitment
- Choices
- Empowerment
- Flexibility
- Partnership

How Long Does it Take for Iron to Build Up?

About a **year** or **20** transfusions

How Do You Check for Iron in the Body?

Blood test: **ferritin** every 3 months

MRI: liver and heart every year

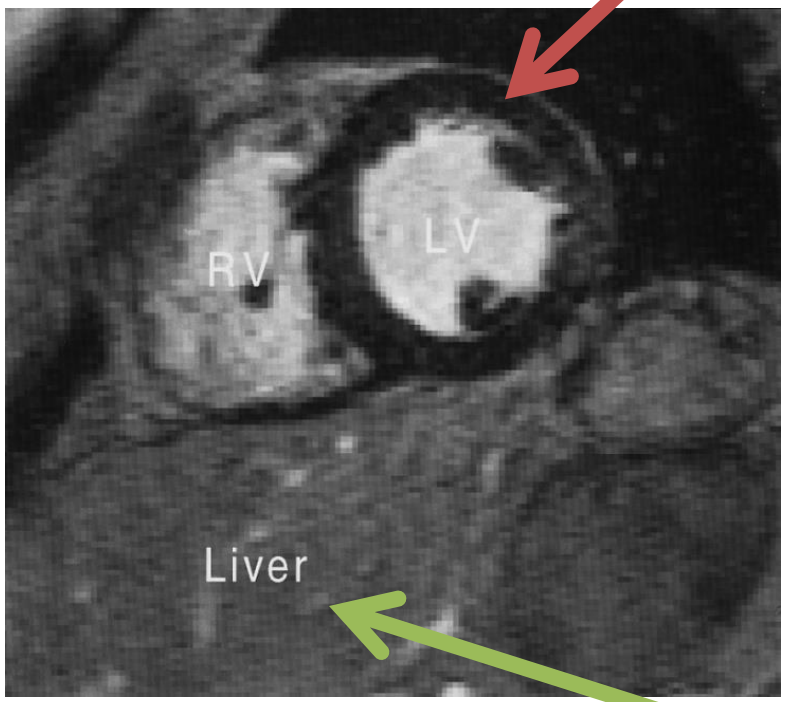
How Does MRI Work?

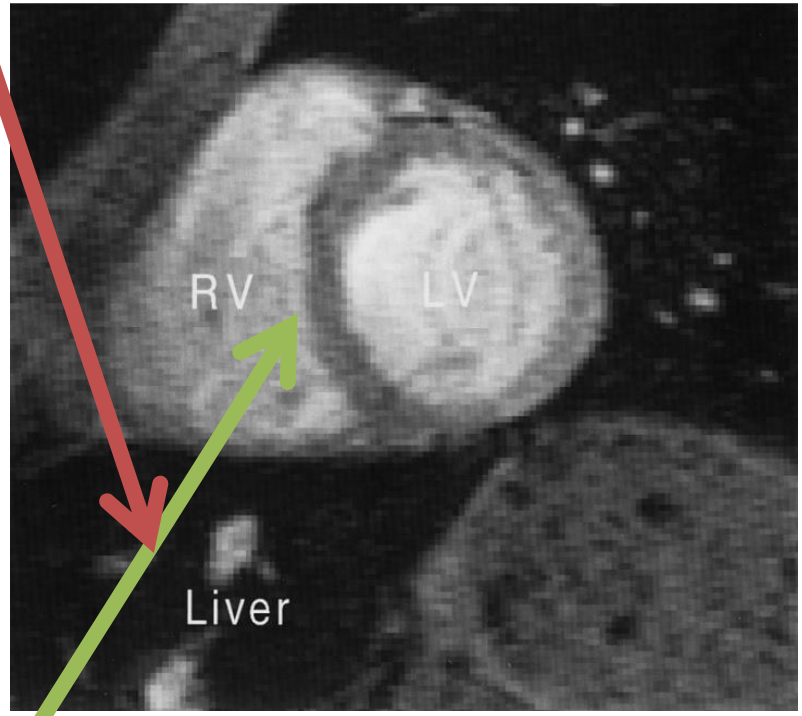
Does not use radiation/xrays

Iron spins faster than other atoms in your body

- measures the spin time which can be converted to iron level

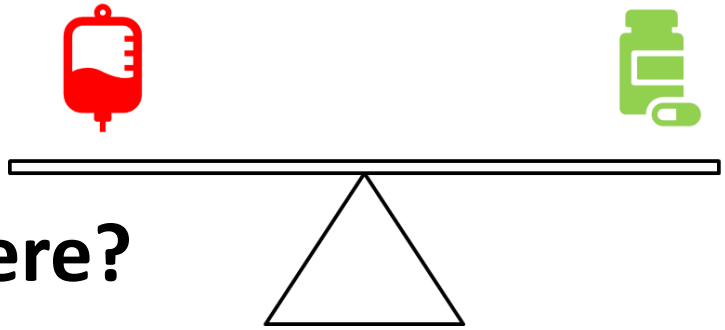
Dark = High (Bad) Iron





Light = Low (Good) Iron Levels

Anderson LJ, et al. Acta Haematol 2006; 115(1–2):106–108.



1. How Much Iron is There?

2. Where is the Iron?

3. What to Take?

1. How Much Iron is There?

	Liver Iron (mg/g)	Heart Iron (T2*)
	**>15**	**<8**
	>7	**10-20**
	<1.8	**>20**

2. Where is the Iron?

Liver scarring, cancer

Thyroid, diabetes, fertility hormones

Heart rhythm and pumping

You are an **individual**

Your needs will **change** over time

But your **target** stays the same

- **reduce** iron levels
- **keep** them low

Deferoxamine (Desferal; DFO)
Deferiprone (Ferriprox; DFP)
Deferasirox (Exjade, JadeNu; DFX)

Deferoxamine (Desferal; DFO)

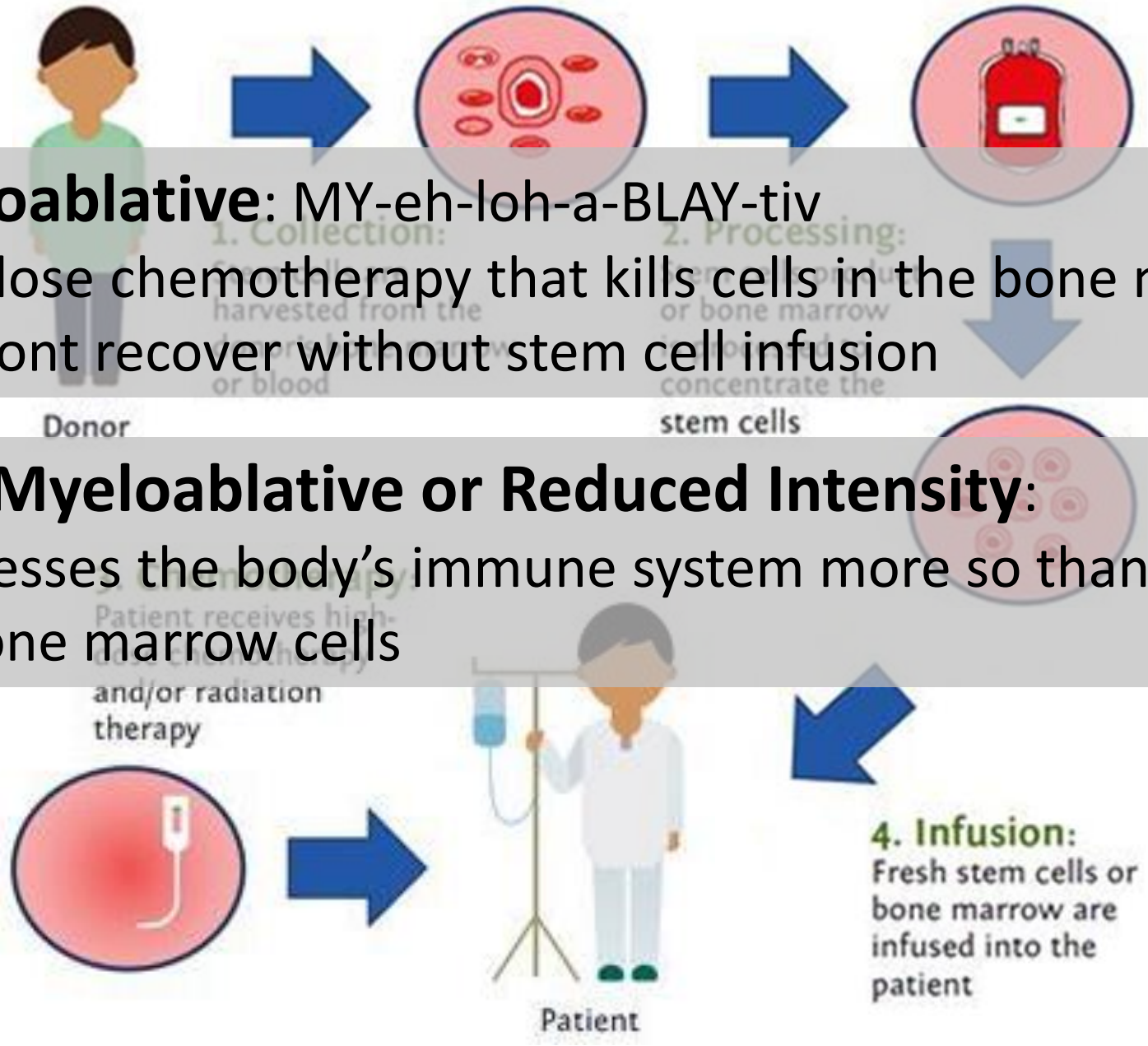
Infusion under the skin overnight or through IV line
Skin reactions, hearing and vision problems
Good for liver iron

Deferiprone (Ferriprox; DFP)

Three times a day, several pills or liquid
Liver irritation, low white blood cells, joint aches
Good for heart iron

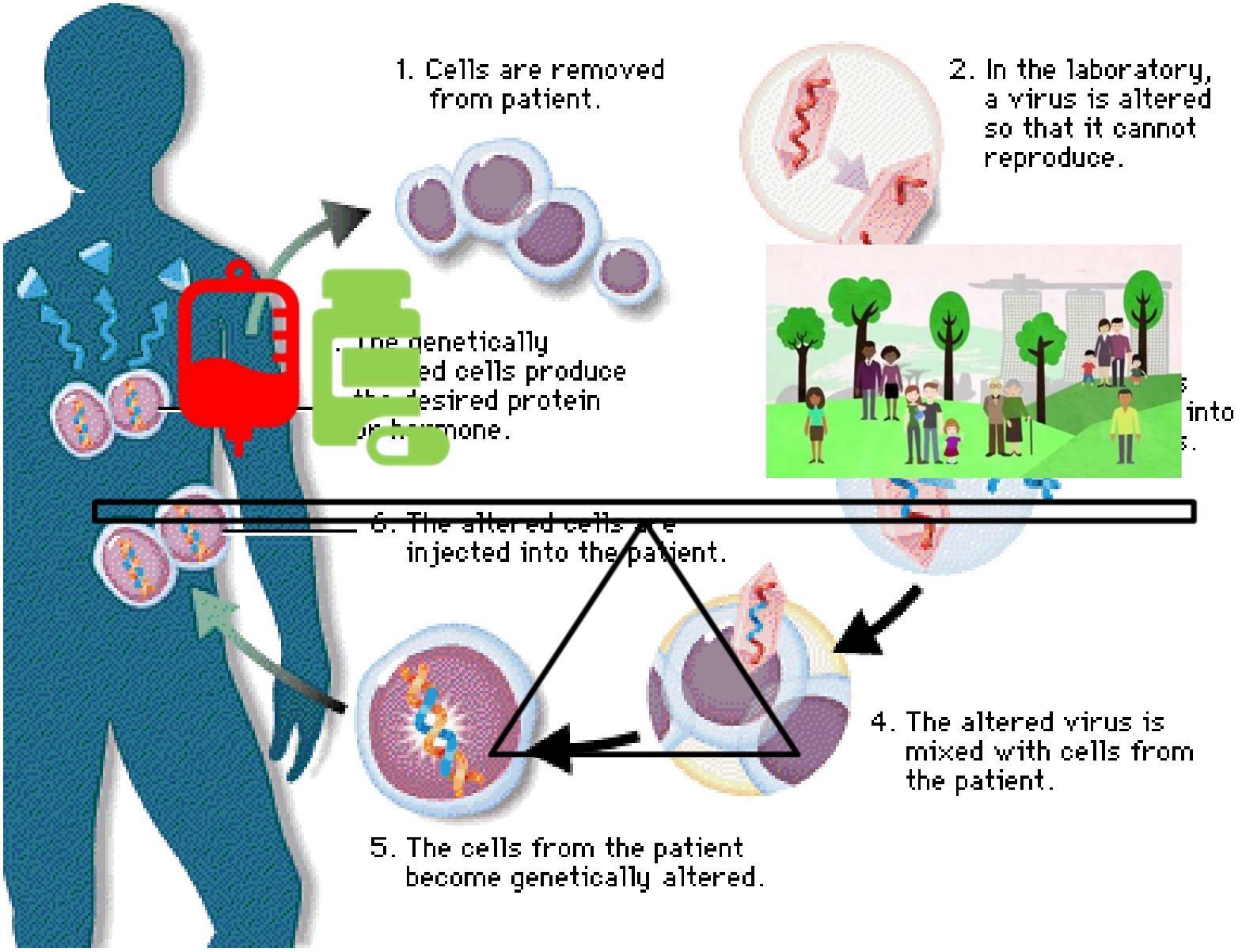
Deferasirox (Exjade, JadeNu; DFX)

Once a day pills or drink
Kidney problems, stomach upset, rash
Good for liver iron



Myeloablative: MY-eh-loh-a-BLAY-tiv
High-dose chemotherapy that kills cells in the bone marrow and wont recover without stem cell infusion

Non-Myeloablative or Reduced Intensity:
Suppresses the body's immune system more so than killing the bone marrow cells


1. Cells are removed from patient.
2. In the laboratory, a virus is altered so that it cannot reproduce.
. The genetically ed cells produce desired protein hormone.
into
6. The altered cells are injected into the patient.
4. The altered virus is mixed with cells from the patient.
5. The cells from the patient become genetically altered.

Who's on YOUR Team?

Hematologist
Nurses/NPs
Physician Assistant
Social Worker
Psychologist
Family & Friends
CAF!

Phlebotomist/IV tech
Blood bank technician
Transfusion medicine MD
Cardiologist, Endocrinologist, Fertility, ObGy
MRI Radiologist
Pharmacist
Insurance company
Teachers
HR department
Politicians


SMALL QUESTIONS
LEAD TO SMALL DISCOVERIES.


BIGGER QUESTIONS
LEAD TO BIGGER DISCOVERIES.


SOME QUESTIONS
ONLY REVEAL DEEPER MYSTERIES.


EVEN IF YOU KNOW WHAT QUESTION TO ASK
THE ANSWER MAY SURPRISE YOU.