

Anemia in CKD Patients

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Why is anemia in
CKD patients
important to me?

Objective:

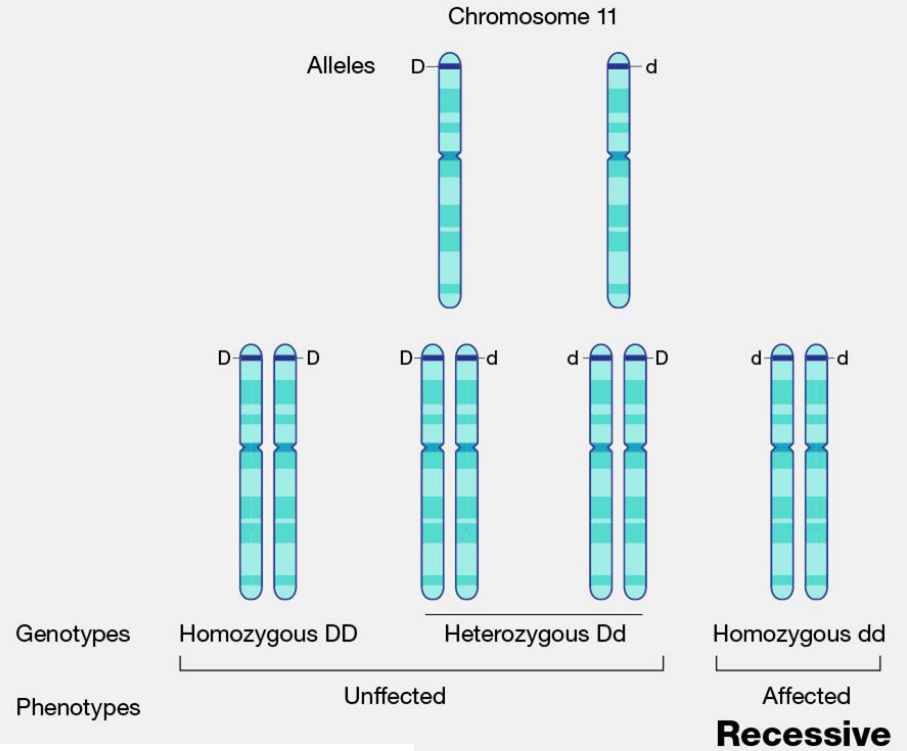
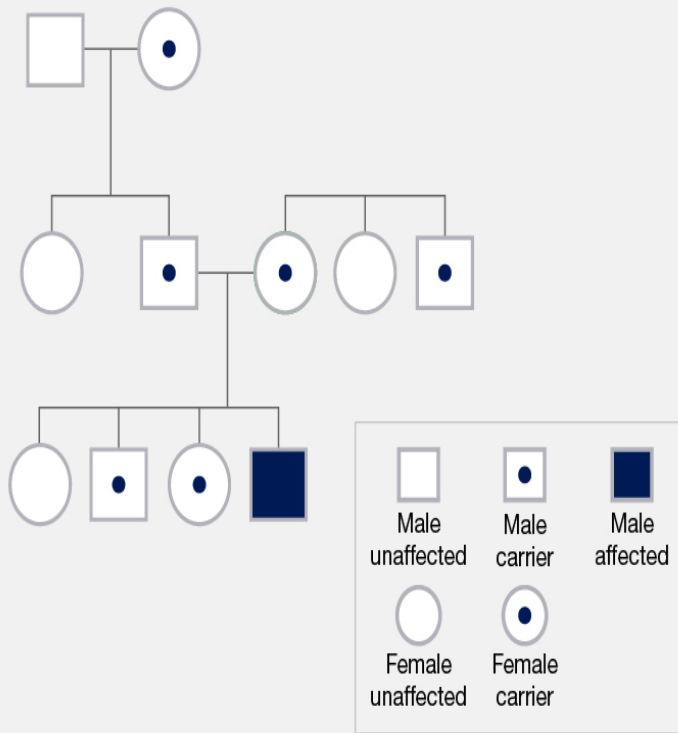
1. Types of anemia
2. What happens when you have anemia?
3. How does anemia affect CKD patients vs an otherwise healthy person?
4. Why are chronic kidney disease patients prone to anemia?
5. How do you treat anemia?
6. A Real World Example
7. Conclusion

Terms to know

- Anemia
 - Autosomal recessive disorder
 - CKD
 - EPO
 - Germline mutation
 - HBB
 - Pica
 - Splice (site) mutation
 - Substitution point mutation
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- TMPRSS6

Types of Anemia

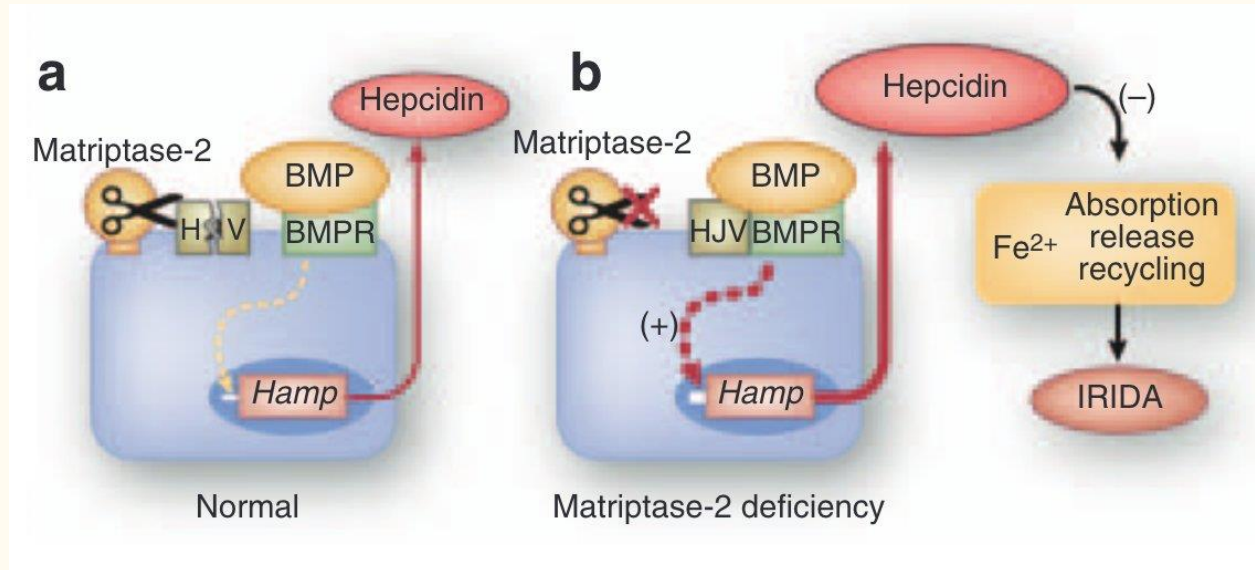
1. Aplastic anemia
2. Iron-refractory iron deficiency anemia (IRIDA)
3. Sickle cell anemia (SCA)
4. Thalassemia
5. Vitamin deficiency anemia



Autosomal recessive disorder

IRIDA

- Caused by *TMPRSS6* gene mutations
- Autosomal recessive disorder
- Germline mutation



SCA

- Caused by HBB gene mutations
- Autosomal recessive disorder
- Substitution point mutation

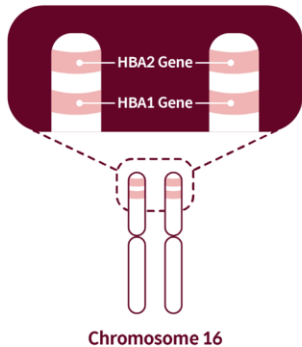
Table 1: Single-Base Mutation Associated with Sickle-Cell Anemia

Sequence for Wild-Type Hemoglobin												
ATG	GTG	CAC	CTG	ACT	CCT	GAG	GAG	AAG	TCT	GCC	GTT	ACT
Start	Val	His	Leu	Thr	Pro	Glu	Glu	Lys	Ser	Ala	Val	Thr
Sequence for Mutant (Sickle-Cell) Hemoglobin												
ATG	GTG	CAC	CTG	ACT	CCT	GTG	GAG	AAG	TCT	GCC	GTT	ACT
Start	Val	His	Leu	Thr	Pro	Val	Glu	Lys	Ser	Ala	Val	Thr

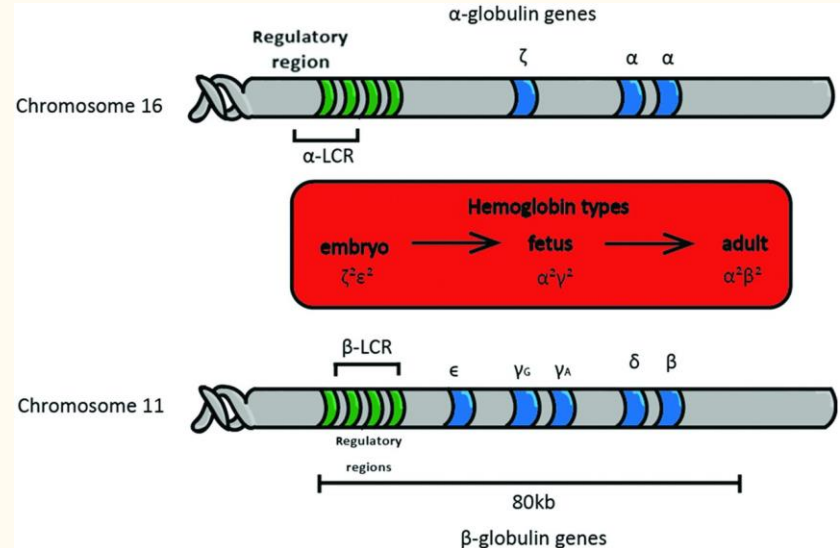
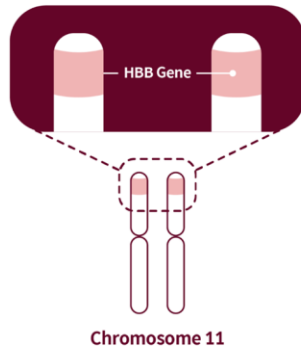
Thalassemia

- Caused by alpha-globin (HBA1 and HBA2) and beta-globin (HBB) genes
- Autosomal recessive disorder
- Splice mutation

ALPHA THALASSEMIA



BETA THALASSEMIA



What happens when you have anemia?

- Fatigue
- Weakness
- Shortness of breath
- Pale or yellowish skin
- Irregular heartbeat
- Dizziness/lightheadedness
- Chest pain
- Cold, numb, or tingling hands and feet
- Headaches
- Difficulty concentrating
- Irritability
- Loss of appetite
- Blue color of the eye whites
- Brittle nails
- Pica symptoms
- Lightheadedness upon standing up
- Sore/inflamed tongue
- Mouth ulcers
- Abnormal menstruation in women
- Loss of libido in men

How does anemia affect CKD patients vs an otherwise healthy person?

In CKD

1. Caused by lack of EPO
2. Patients experience anemia more frequently
3. Often require EPO stimulating agents

In otherwise healthy people

1. Caused by various factors
2. Patients experience anemia less frequently or chronically if by genetic mutation
3. Often can be treated with any number of treatments and quicker than in CKD

Why are CKD patients prone to anemia?

- Kidney damage
 - Waste build up
 - Lesser EPO production
 - Fewer RBC production
 - Shorter RBC life
 - Lack of necessary nutrients
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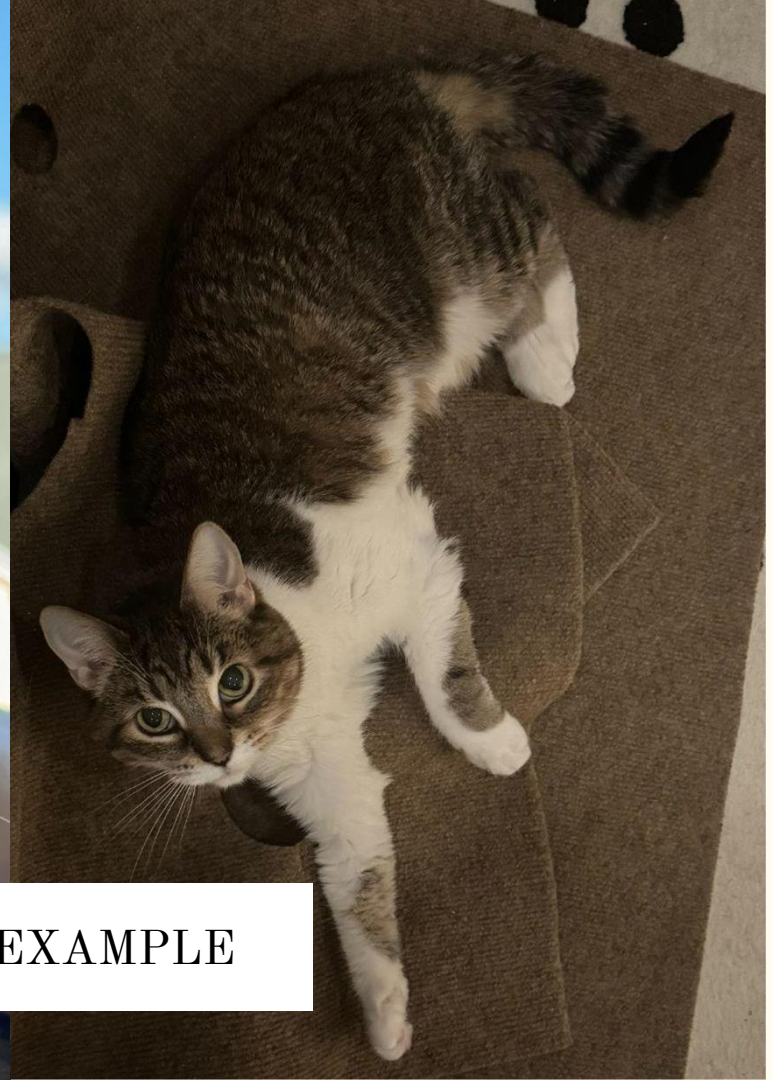
How do you treat anemia?

Tests

- CBC
- Hemoglobin test
- Hematocrit test
- Peripheral blood smear
- Reticulocyte count

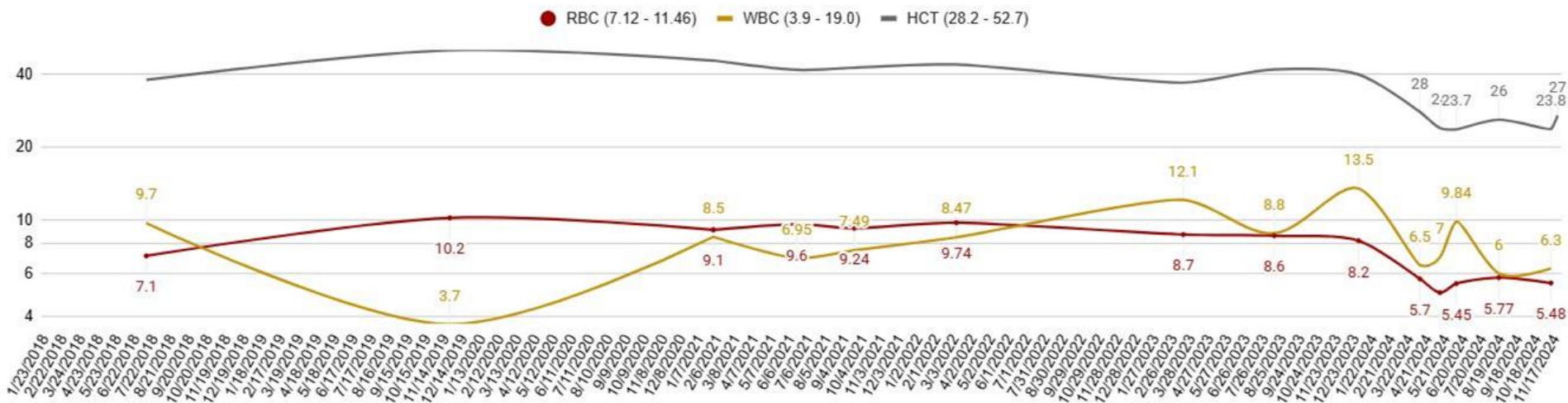
Treatments

- Iron supplements
- Folate (B9)
- B12 supplements
- Erythropoietin
- Corticosteroids
- Blood transfusion
- Bone marrow transplant



A REAL WORLD EXAMPLE

Anemia



Effects of Varenzin, an erythropoietin treatment on Alison

Conclusion

- Anemia as result of mutation can be caused by 3 possible types of gene mutations
- Genetic mutation anemias are autosomal recessive disorders
- TMPRSS6, HBB, HBA1, or HBA2 genes
- CKD patients experience anemia often

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