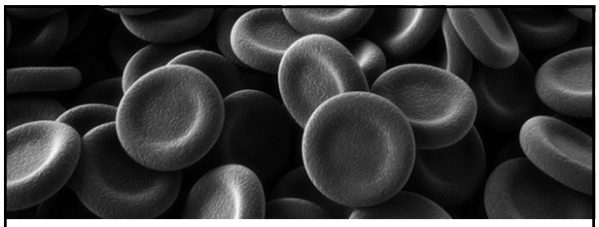

Hemoglobinopathies in Pregnancy

Emily Parkhurst, MS, LCGC
Kaiser West Los Angeles

November 2017

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Quiz

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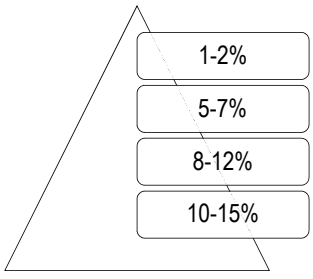
What percentage of the world's population is a carrier of a hemoglobinopathy?



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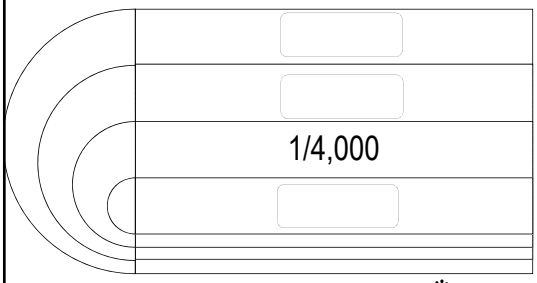
How common are babies with hereditary anemias born in California?



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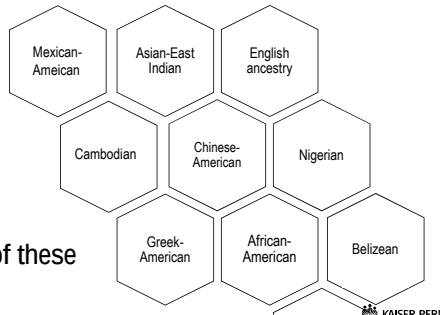
Who have I counseled patients about being a carrier of a hemglobinopathy?



All of these

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Why are hemoglobinopaties important?

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Genetic disorders of Hemoglobin

- The World Health Organization estimates that about 5-7% of world's population carries a clinically significant hemoglobin variant.[1]
- The rate of occurrence of hemoglobinopathies in California is more than 1 in 4,000 births.[2]
- More than 400 clinically abnormal hemoglobins have been identified and about half are clinically significant

1. World Health Organization, Sickle-cell disease and other haemoglobin disorders, Jan 2011
2. California Department of Health Care Services, Systems of Care Division Child Health and Disability Prevention Program, Health Assessment Guidelines March 2016

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Hemoglobinopathies in Pregnancy

- Genetics
- Different Types of Conditions
- Clinical Recommendations
- Counseling

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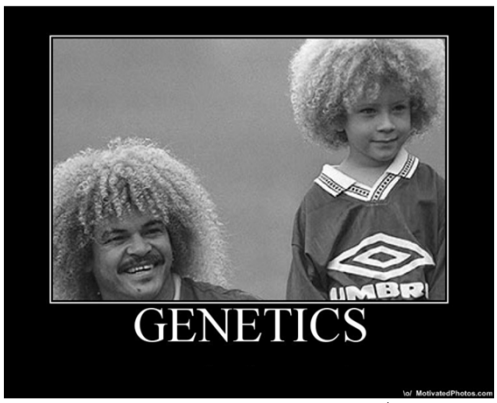
Refer to Genetics

When in doubt, send a Dr. Advice to genetics.



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How are hemoglobinopathies passed in families?

Autosomal Recessive

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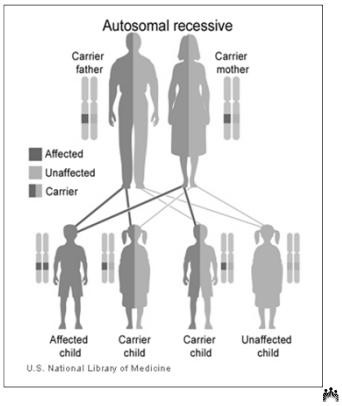

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Autosomal recessive

Heterozygotes

- Hb AS = Sickle cell trait
- Hb AC = Hemoglobin C trait

Homozygotes

- Hb SS = sickle cell anemia
- Hb SC = sickle cell disease

Sickle cell is most common people of African ancestry, but also seen in people of Latin-American, Mediterranean and Asian East Indian.

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Why are hemoglobinpathies more common in certain populations?

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Sickle Cell Distribution

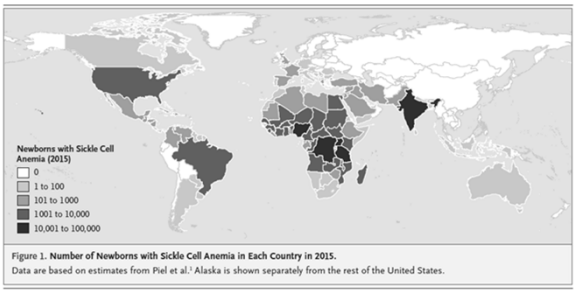

Figure 1. **Number of Newborns with Sickle Cell Anemia in Each Country in 2015.**
Data are based on estimates from Piel et al.[3] Alaska is shown separately from the rest of the United States.

Longo, D. Sickle Cell Disease. NEJM 2017

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Heterozygote Advantage

Carriers of sickle cell trait have red cells that are inhospitable to the malaria organism

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Different Types of Inherited Anemias

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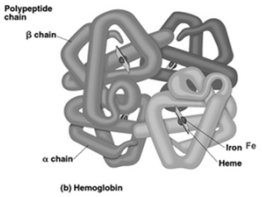
Hemoglobin molecule

- Hb A: $\alpha_2\beta_2$
  - A for "adult" hemoglobin
- Beta globin gene mutations
  - Hb S, Hb C, Hb E, etc
  - Beta thalassemia
- Alpha globin gene mutations
  - Alpha thalassemia
  - Hb Constant Spring



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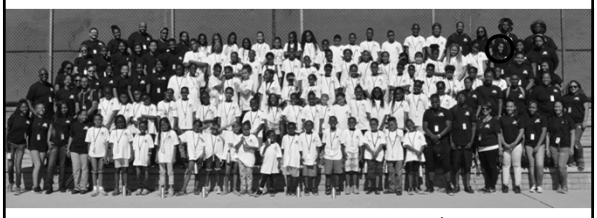
What is a sickle?

SICKLE CELL DISEASE
FOUNDATION OF CALIFORNIA

Inspiring DREAMS Camp Crescent Moon



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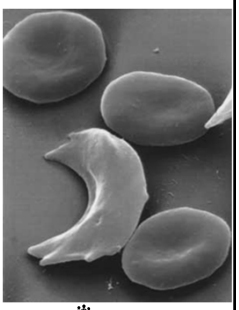
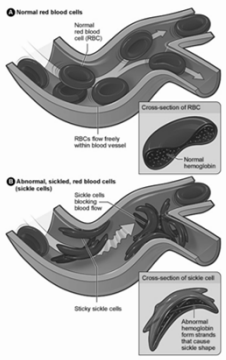
Sickle Cell Pathophysiology

- In deoxygenated blood, sickle red cells are only 1/5 as soluble as normal hemoglobin.
- Under conditions of low oxygen tension, Hb S molecules aggregate into rod shape polymers that distort the round shape of a red cell.
- These misshapen 'sickle' cells are less flexible than normal and cannot squeeze single file through capillaries, blocking blood flow and causing ischemia.



KAISER PERMANENTE



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Sickle cell trait

CBC NO DIFF
Status: **Final result** Visible to patient: **kp.org** Next appt: **None** Dx: **SUPERVISION NORMAL FIRST PREGNANCY, F...**

Newer results are available. Click to view them now.

	Ref Range	1/13/15 10:03 AM
WBC'S AUTO	4.0 - 11.0 x1000/mcL	4.4
RBC, AUTO	4.20 - 5.40 Mill/mcL	4.47
HGB	12.0 - 16.0 g/dL	13.2
HCT, AUTO	37.0 - 47.0 %	39.0
MCV	81.0 - 99.0 fL	87.2
MCH	27.0 - 35.0 pg/cell	29.5
MCHC	32.0 - 37.0 g/dL	33.8
RDW, BLOOD	11.5 - 14.5 %	13.1
PLATELETS, AUTOMATED COUNT	130 - 400 x1000/mcL	157

HEMOGLOBIN EVALUATION
Status: **Final result** Visible to patient: **Not Released** Dx: **SUPERVISION NORMAL FIRST PREGNANCY, F...**

	Ref Range	1/13/15 10:03 AM	1/13/15 10:03 AM
HEMOGLOBIN F %, HPLC	<=2.0 %	0.2	
HEMOGLOBIN PHENOTYPE, BLOOD		See Confrm Test	AS (A)

Narrative
RMS ACCN: 553357320

Specimen Collected: 01/13/15 10:03 AM Last Resulted: 01/15/15 1:26 PM Lab Flowsheet Order Details View Encounter Lab

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Sickle Cell Trait

Usually does not cause any serious health problems

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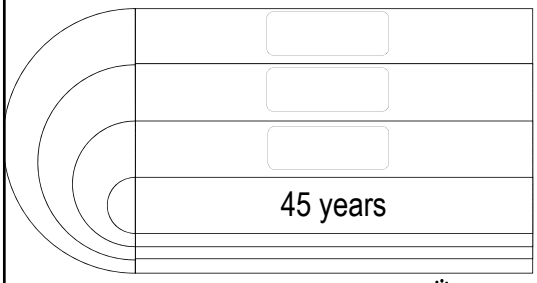
What is the life expectancy for sickle cell anemia?



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Sickle Cell Anemia

a multisystem disorder

- Pain crises
  - Due to ischemic tissue from obstructed blood flow, hypoxia and acidosis
  - May last several days to weeks
  - Triggered by fever, dehydration, cold, stress
- Infection
  - Immune system is compromised because the spleen is not functioning properly or missing.
  - Flu, sepsis, meningitis, pneumonia

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Sickle Cell Anemia
a multisystem disorder

- Stroke
  - Ischemic or hemorrhagic lesion in a specific vascular area
- Retinopathy
  - Retinal hemorrhage if vessels in eye are blocked
- Acute chest syndrome
  - Due to acute pulmonary infarction – pneumonia like symptoms
- Splenic Sequestration (enlarged)

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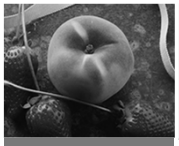
Prevention for Sickle Cell Anemia



Routine care	Healthy Diet	Medications
Vaccinations	Hydradtion Folic Acid supplements	Anti-inflamatory agents Prophylactic antibiotics

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Treatment for Sickle Cell Anemia

Blood transfusions
Anemia treatment
Stroke prevention
Chelation therapy
Pain management

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What are other hemoglobin variants besides sickle cell?

What are other hemoglobin variants besides sickle cell?

- Hb C
- Hb E
- Hb D
- Hb O

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Hb HaSharon

Hb Pisa

Hb Hyde Park

Hb S - Sickle

Hb G-Philadelphia

Hb Hope

Hb Tak

More than 400 abnormal hemoglobins have been described

Hb N-Baltimore

Hb Kansas

Hb E

Hb Hammersmith

Hb D - Punjab

Hb Lepore

Hb O - Arab

Hb C

Hb M

Hb Kempsey

Hb Korle-Bu

Hb H

Hb Gun Hill

Hb Constant Spring

Hb Miyada

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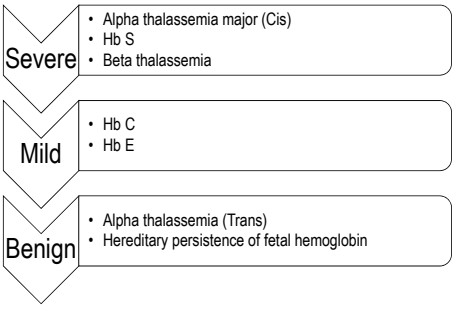
Spectrum of severity



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Which population has the highest carrier frequency?

Carrier frequencies

Ethnicity	β thal	α thal	Hb S	Hb C	Other Hb
Mediterranean	1/20-30	1/40	1/40	rare	D, G, lepore
African American	1/75	1/30 trans	1/12	1/50	O,D
African Caribbean	1/50-75	1/30 trans	1/12	1/30	O,D
West-African	1/50	1/30 trans	1/6	1/20-30	O,D
Hispanic Caribbean	1/75	Variable	1/30	rare	Variable
Hispanic Latino	1/30-50	Variable	1/30-200	rare	J,E
Asian	1/50	1/20 cis	Rare	rare	E
Southeast Asian	1/30	1/20 cis	rare	rare	E 1/2-1/3
Asian Indian	1/30-150	Variable	1/50	rare	D, O, E
Middle Eastern	1/50	Variable	1/50	rare	D, O, E

March of Dimes Genetic Screening Pocket Facts

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Hemoglobin E

- Structural variant resulting in decreased synthesis of hemoglobin.
- Most common structural hemoglobin abnormality in the world
  - About 1/10 Southeast Asians carry Hb E
  - Up to 1/3 carries Hb E in parts of Laos, Cambodia, Thailand
  - Up to 1/2 in parts of Northern India
- Homozygous EE mild anemia, often asymptomatic
- Only a risk for disease when combined with β thal trait

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Hb E trait

HEMOGLOBIN VARIANT CONFIRMATION, ELECTROPHORESIS

Status: Final result Visible to patient: No (Not Released) Next appt: 10/23/2017 at 08:00 AM in Obstetrics, Gynecology (KELLIE

		Ref Range & Units	9/13/17 9:11 AM
HEMOGLOBIN A,BLD,QN, ELECTROPHORESIS		%	71.3
HEMOGLOBIN A2, TOTAL, BLOOD, EP		%	3.1
Comments:			
Hb A2 reference ranges for patients with select hemoglobin variants:			
Hgb S 2.2-3.9%			
Hgb D 2.0-3.6%			
Hgb E 2.8-4.5%			
Hb A2 levels are not a specific indicator of an underlying beta-thalassemia in the presence of these hemoglobin variants.			
HGB E %, BLOOD, ELECTROPHORESIS		%	25.6
HEMOGLOBIN PHENOTYPE, BLOOD		AA	AE !
HGB PHENOTYPE INTERPRETATION, BLOOD, ELECTROPHORESIS			Heteroz...
Heterozygous Hb E (Hb E Trait). This is a clinically benign condition which produces no anemia. Hematology consultation is not necessary, but reproductive counseling may be advisable. Anemia may indicate the presence of an underlying thalassemia.			

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Hemoglobin C

- Similar to Hb S
  - (the same DNA position as the sickle cell mutation but different amino acid substitution)
  - Compound heterozygotes (Hb SC) are affected with "SC disease" which varies, but is often a milder anemia than sickle cell disease.
- Oxygenated Hb C tends to crystallize, leading to less flexible red cells and mild hemolysis.
- About 1/30-1/50 African Americans carries Hb C

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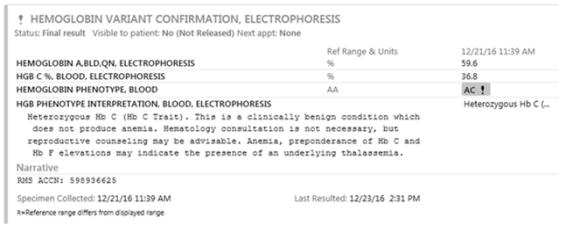
Hb C trait



HEMOGLOBIN VARIANT CONFIRMATION, ELECTROPHORESIS

Status: Final result Visible to patient: No (Not Released) Next appt: None

	Ref Range & Units	12/21/16 11:39 AM
HEMOGLOBIN A,BLD,QN, ELECTROPHORESIS	%	59.6
HGB C %, BLOOD, ELECTROPHORESIS	%	36.8
HEMOGLOBIN PHENOTYPE, BLOOD	AA	AC !
HGB PHENOTYPE INTERPRETATION, BLOOD, ELECTROPHORESIS		Heterozygous Hb C (...

Heterozygous Hb C (Hb C Trait). This is a clinically benign condition which does not produce anemia. Hematology consultation is not necessary, but reproductive counseling may be advisable. Anemia, preponderance of Hb C and Hb F elevations may indicate the presence of an underlying thalassemia.

Narrative

RMS ACCN: 598936625

Specimen Collected: 12/21/16 11:39 AM Last Resulted: 12/23/16 2:31 PM

*=Reference range differs from displayed range

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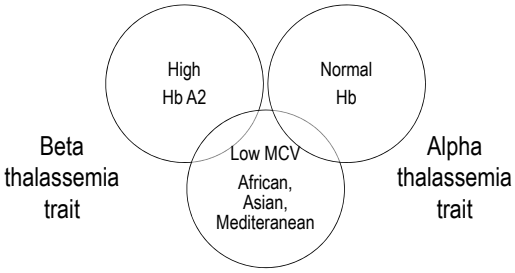
Refer to Genetics

When in doubt, send a Dr. Advice to genetics.

What's the difference between alpha and beta thalassemia?

How to tell thalassemia carriers apart?



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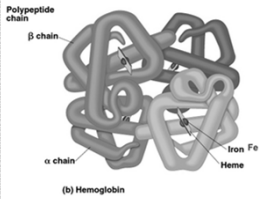
Normal Hemoglobin

- Normal levels
  - MCV 80-100 fL
  - Hb A > 95%
  - Hb A2 < 3.5%
  - Hb F <2 %
- Hb A: $\alpha_2\beta_2$
- Hb A2: $\alpha_2\delta_2$
- Hb F: $\alpha_2\gamma_2$



KAISER PERMANENTE

Beta thalassemia trait

HEMOGLOBIN FRACTIONATION, CHROMATOGRAPHY

Status: Final result Visible to patient: No (Not Released) Next appt: Today at 09:30 AM in Genetics (EMILY S PARKHURST) Dx: ANTENATAL SCREENING

	Ref Range & Units	9/14/17 8:14 AM	Flag
HEMOGLOBIN A %, HPLC	%	91.5	
HEMOGLOBIN A2 %, HPLC	1.5 - 3.5 %	5.6 ^	H
HGB A2 INTERPRETATION, BLOOD, HPLC		Hb A2 is elevated. In the context of microcytic anemia, these results are usually seen in patients with beta thalassemia. Hematology consultation is not necessary, but reproductive counseling may be advisable.	
HEMOGLOBIN F %, HPLC	<=2.0 %	1.7	

CBC NO DIFFERENTIAL

Status: Final result Visible to patient: No (Not Released) Next appt: Today at 09:30 AM in Genetics (EMILY S PARKHURST) Dx: ANTENATAL SCREENING

	Ref Range & Units	9/14/17 8:14 AM	Flag
WBC'S AUTO	4.0 - 11.0 x1000/mcL	5.4	
RBC, AUTO	4.20 - 5.40 Mil/mcL	5.53 ^	H
HGB	12.0 - 16.0 g/dL	10.9 v	L
HCT, AUTO	37.0 - 47.0 %	34.8 v	L
MCV	81.0 - 99.0 fL	63.0 v	L
MCH	27.0 - 35.0 pg/cell	19.7 v	L
MCHC	32.0 - 37.0 g/dL	31.2 v	L
RDW, BLOOD	11.5 - 14.5 %	15.5 ^	H
PLATELETS, AUTOMATED COUNT	130 - 400 x1000/mcL	200	

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Beta thalassemia

- Decreased production of β chains, increased α chains
  - High levels of α chains damage bone marrow and decrease erythropoesis
  - Higher levels of Hb A2
- Most common in people of Mediterranean, North African, and South-East Asian ancestry
- Variable severity depending on mutation and level of β chain production
  - β^0 thalassemia – no Hb A produced, more severe
  - β^+ thalassemia – some Hb A produced, mild
- Similar symptoms and treatment as sickle cell disease
- Also called Cooley's anemia

Genetic Home Reference: Your guide to understanding genetic conditions. (2012)
Weatherall DJ1. Phenotype-genotype relationships in monogenic disease: lessons from the thalassemias. Nat Rev Genet. 2001

KAISER PERMANENTE

Alpha thalassemia trait

HEMOGLOBIN FRACTIONATION, CHROMATOGRAPHY

Status: Final result Visible to patient: No (Not Released) Next appt: 09/29/2017 at 03:00 PM in Radiation Oncology (MICHAEL RAYMOND GIRVIGIAN MD, M.D.) Dx:

	Ref Range & Units	9/11/17 12:28 PM
HEMOGLOBIN A %, HPLC	%	95.7
HEMOGLOBIN A2 %, HPLC	1.5 - 3.5 %	2.7
HEMOGLOBIN F %, HPLC	<=2.0 %	0.4
HEMOGLOBIN PHENOTYPE, BLOOD		AA
HGB PHENOTYPE INTERPRETATION, BLOOD, HPLC		No abnormality detected. If the patient is not anemic or has nutritionally-responsive anemia, no further testing is needed. In the

† CBC NO DIFFERENTIAL

Status: Final result Visible to patient: Yes (kp.org) Next appt: 09/29/2017 at 03:00 PM in Radiation Oncology (MICHAEL RAYMOND GIRVIGIAN MD, M.D.) Dx: ROUTINE ADULT HEALTH CHECK UP EXAM

Notes Recorded by Tuason, Angelica S (R.N.), R.N. on 8/8/2017 at 10:14 AM
Letter sent

Notes Recorded by Zheng, Xiaona (M.D.), M.D. on 8/7/2017 at 6:48 PM
Call or letter ↑ Blood test is normal; no more anemia

	Ref Range & Units	8/7/17 9:59 AM	1/13/16 12:30 PM	12/24/15 5:55 AM	12/25/15 5:58 AM	12/19/15 8:39 AM
WBC'S AUTO	4.0 - 11.0 x1000/mcL	5.5	7.8	15.6 ^	13.0 ^	5.2
RBC, AUTO	4.20 - 5.40 MIL/mcL	5.65 ^	5.38	4.31	4.41	5.28
HGB	12.0 - 16.0 g/dL	12.5	11.4 v	8.7 v	8.4 v	10.7 v
HCT, AUTO	37.0 - 47.0 %	39.0	36.5 v	29.0 v	30.7 v	35.6 v
MCV	81.0 - 99.0 fL	69.1 v	67.9 v	67.2 v	69.7 v	67.4 v
MCH	27.0 - 33.0 pg/cell	22.1 v	21.2 v	20.2 v	20.3 v	20.2 v
MCHC	31.0 - 37.0 g/dL	32.1	31.2 v	30.0 v	29.4 v	30.0 v
RDW, BLOOD	11.5 - 14.5 %	14.7 ^	18.9 ^	18.8 ^	18.4 ^	17.6 ^

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Alpha thalassemia trait

FERRITIN

Status: Final result Visible to patient: Yes (kp.org) Next appt: 09/29/2017 at 03:00 PM in Radiation Oncology (MICHAEL RAYMOND GIRVIGIAN MD, M.D.) Dx: TH

	Ref Range & Units	9/11/17 12:28 PM
FERRITIN	13 - 126 ng/mL	34

Comments: Iron therapy of anemic chronic kidney disease and dialysis patients may be adjusted to produce serum ferritin levels between 100 and 1200 ng/ml.

† IRON AND TIBC

Status: Final result Visible to patient: Yes (kp.org) Next appt: 09/29/2017 at 03:00 PM in Radiation Oncology (MICHAEL RAYMOND GIRVIGIAN MD, M

	Ref Range & Units	9/11/17 12:28 PM
IRON	37 - 145 mcg/dL	177 ^
TOTAL IRON BINDING CAPACITY	250 - 450 mcg/dL	361
IRON SAT	20 - 50 %	49

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Previously normal MCV

CBC W AUTOMATED DIFFERENTIAL

Status: **Final result** Visible to patient: **No (Not Released)** Next appt: 09/19/2017 at 10:00 AM in Genetics (EMILY S PARKHURST) Dx: ABNL VAGINAL BLEEDING

Newer results are available. Click to view them now.

	Ref Range & Units	11/13/15 8:35 AM	10/12/14 5:14 AM	10/11/14 7:54 AM	7/17/14 11:41 AM
WBC'S AUTO	4.0 - 11.0 x1000/mcL	6.1	9.7	8.1	9.4
RBC, AUTO	4.20 - 5.40 Mil/mcL	3.30	2.87	3.90	3.79
HGB	12.0 - 16.0 g/dL	6.1 !!	7.6	10.1	10.6
Comments:					
Critical Result called to: N.ONTIVEROS RN Date/Time: 11/13/2015 10:27:02 AM PST_ By:KZ53633_ Read back of verbal test result(s) confirmed					
HCT, AUTO	37.0 - 47.0 %	20.9	22.9	30.6	31.1
MCV	81.0 - 99.0 fL	63.3	80.0	78.4	82.1
MCH	27.0 - 35.0 pg/cell	18.4	26.6	25.9	28.0
MCHC	32.0 - 37.0 g/dL	29.0	33.3	33.1	34.1
RDW, BLOOD	11.5 - 14.5 %	23.1	14.5	13.9	13.5
PLATELETS, AUTOMATED COUNT	130 - 400 x1000/mcL	257	128	135	195

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α thalassemia

There are two distinct types of α thalassemia mutations

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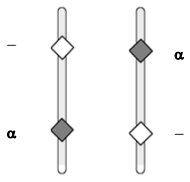
Weatherall DJ1. Phenotype-genotype relationships in monogenic disease: lessons from the thalassemias. Nat Rev Genet. 2001

Alpha Thalassemia Trait:

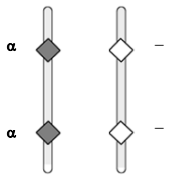
Two Forms of Inheritance

One working gene on each chromosome



Trans Arrangement
African/Mediterranean ancestry

Working genes on same chromosome



Cis Arrangement
Asian ancestry

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α thalassemia

There are four alpha globin genes in total.

- Normal (aa / aa)
- "Silent" carriers have 3 functional α genes (-a / aa)
- Carriers have 2 functional α genes (-a / -a)
  or (-- / aa)
- Hb H disease due to only α gene (-- / -a).
- Hydrops fetalis/ Hb Barts no α genes (-- / --).

The lethal form of alpha thalassemia from hydrops fetalis due to two cis mutations, which is associated with **Asian** ancestry.

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How to tell thalassemia carriers apart?

Low MCV

Normal Hb	↑Hb A2
• Iron deficiency • Alpha thal trait	• Beta thal trait

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Refer to Genetics

When in doubt, send a Dr. Advice to genetics.

Benign hemoglobin traits

What are some benign hemoglobin variants?

- Hb A2' (or delta chain variant)
- Elevated Hb F

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Hb A2' /delta chain variant

- The A2' (also called A2 prime or a delta-chain variant) is not a hemoglobinopathy trait and does not affect health

HEMOGLOBIN VARIANT CONFIRMATION, ELECTROPHORESIS

Status: Final result Visible to patient: No (Not Released) Next appt: 10/09/2017 at 02:50 PM in Obstetrics, Gynecology (NICOLE NAM

	Ref Range & Units	8/25/17 2:52 PM
HEMOGLOBIN PHENOTYPE, BLOOD	AA	A2P !
HGB PHENOTYPE INTERPRETATION, BLOOD, ELECTROPHORESIS		Hb A2' ...

Hb A2' (Also known as Hb B2). This patient has a delta-chain variant, most common among people of African or Sicilian descent. This is of no known hematologic consequence. Both Hb A2 and Hb A2' are included in the reported Hb A2 percentage.

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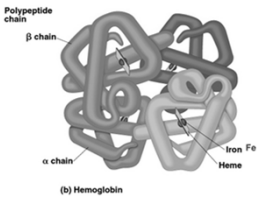
Normal Hemoglobin

- Normal levels
  - MCV 80-100 fL
  - Hb A > 95%
  - Hb A2 < 3.5%
  - Hb F <2 %
- Hb A: $\alpha_2\beta_2$
- Hb A2: $\alpha_2\delta_2$
- Hb F: $\alpha_2\gamma_2$



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Hereditary Persistence of Fetal Hemoglobin

- Individuals with such deletions may have hereditary persistence of fetal hemoglobin (HPFH) with up to 35% Hb F in adults; this is a benign condition.
  - Hydroxyurea uses this mechanism to increase Hb F in blood to treat patients with sickle cell anemia and beta thalassemia
- A slight elevation in Hb F (3-5%) is not a hemoglobinopathy trait and is naturally associated with pregnancy.

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Hereditary Persistence of Fetal Hemoglobin

HEMOGLOBIN EVALUATION — Order

Status: Final result Visible to patient: No (Not Released) Next appt: None

	Ref Range & Units	8/23/13 4:40 PM	Flag
HEMOGLOBIN A %, HPLC	%	93.4	
HEMOGLOBIN A2 %, HPLC	1.5 - 3.5 %	2.8	
HEMOGLOBIN F %, HPLC	<=2.0 %	2.1 ^	H
HEMOGLOBIN PHENOTYPE, BLOOD		See above	

HEMOGLOBIN EVALUATION

Status: Final result Visible to patient: No (Not Released) Next appt: None Dic PRENATAL INTAKE INTERVIEW

	Ref Range & Units	8/7/15 4:28 PM	Flag
HEMOGLOBIN A %, HPLC	%	90.5	
HEMOGLOBIN A2 %, HPLC	1.5 - 3.5 %	2.4	
HEMOGLOBIN F %, HPLC	<=2.0 %	6.8 ^	H
HGB F INTERPRETATION, BLOOD, HPLC		Elevated Hb F. This pattern can occur in the presence of hereditary persistence of fetal hemoglobin (HPFH) or beta-thalassemia. Isolated HbF elevation can also occur with pregnancy, myeloproliferative disorders, and marrow failure. Up to 10% Hb F can be found in normal patients 1-2 years of age, but Hb F rarely exceeds 2% thereafter.	
HEMOGLOBIN PHENOTYPE, BLOOD		SEE ABOVE	

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Hereditary Persistence of Fetal Hemoglobin

HEMOGLOBIN EVALUATION

Status: Final result Visible to patient: No (Not Released) Next appt: None

Notes Recorded by Gogia, Raveen Kaur (M.D.) on 5/23/2012 at 9:30 AM
Ref genetics sent.

	Ref Range & Units	5/17/12 10:30 AM
HEMOGLOBIN F %, HPLC	<2.0 %	26.0 ^
Comments: Elevated Hb F. This pattern can occur in the presence of hereditary persistence of fetal hemoglobin (HPFH) or beta-thalassemia. Isolated Hb F elevation can also occur with pregnancy, myeloproliferative disorders, and marrow failure. Up to 10% Hb F can be found in normal patients 1-2 years of age, but Hb F rarely exceeds 2% thereafter.		
HEMOGLOBIN A2 %, HPLC	1.5 - 3.5 %	1.9
HEMOGLOBIN A %, HPLC	%	71.8

Specimen Collected: 05/17/12 10:30 AM Last Resulted: 05/21/12 1:39 PM

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Clinical Recommendations

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Clinical Recommendations

CBC for all pregnant women or women planning pregnancy. Hemoglobin electrophoresis in those of certain ethnicities (African, Mediterranean, Middle Eastern, Southeast Asian, West Indian)

CBC and Hemoglobin evaluation for all pregnant women, regardless of ancestry

ACOG Committee Opinion No. 691: Carrier Screening for Genetic Conditions. Mar 2017
Regional SCAL Kaiser Permantete Genetics policy. April 2016

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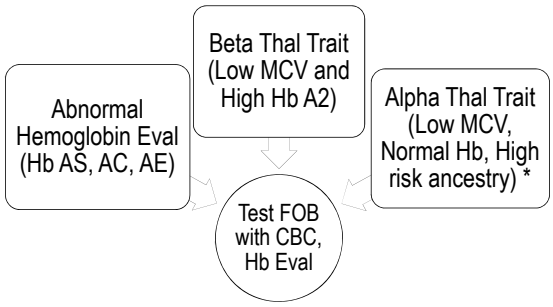
Hemoglobinopathy algorithm

Complete blood count (CBC) and hemoglobin evaluation



*Also serum iron studies as indicated

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Testing the partner/ FOB

Kaiser will cover hemoglobinopathy screening for non-member FOBs

- If a pregnant patient is a carrier of a hemoglobinopathy, testing for the her partner (FOB) is a covered benefit and part of her prenatal care.
- There are no copays or cost sharing for him.
  - If the partner has an MRN from the past, we use that
  - Call MRN services to create a temporary Kaiser number for him

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Refer to Genetics

When in doubt, send a Dr. Advice to genetics.

Counseling

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Health Care Disparities

Recent studies have shown that despite the improvements in the overall health of the country, racial and ethnic minorities experience a lower quality of health care—they are less likely to receive routine medical care and face higher rates of morbidity and mortality than nonminorities.

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Health Care Disparities

Major findings on racial and ethnic gaps in health care

- Disparities in health care exist and are associated with worse health outcomes.
- Health care disparities occur in the context of broader inequality.
- There are many sources across health systems, providers, patients and managers that contribute to disparities.
- Bias, stereotyping, prejudice and clinical uncertainty contribute to disparities.
- A small number of studies suggest that racial and ethnic minority patients are more likely to refuse treatment.

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Counseling issues

Medical literacy

- Use words patients will understand
- Many patients are visual learners
  - Show they the lab results
  - Draw a picture or a Punnett square
- Reinforce that the patient inherited the condition from a parent and that other family members (siblings) may also be carriers.
- Normalize/ reduce stigma
  - Emphasize that every populations has *some* genetic condition

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Counseling issues

Reluctant partners

- Validate concerns
  - Fear of needles, mistrust of doctors, health care disparities are real
  - Reassurance
- Review the facts
  - People who carry the trait feel fine. The only way to know is from a blood test.
- Encourage teamwork
  - It is only fair that if the woman had to get all these blood tests, he can do some too
  - Ultimately, it is his choice and he can refuse testing

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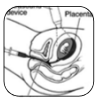
Prenatal Diagnosis



CVS: 10-13 weeks

Aminocentesis: 15-20 weeks

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Counseling issues

Prenatal diagnosis

- CVS and amniocentesis is indicated when both parents are carriers
  - If the father is unavailable for testing, prenatal diagnosis is also indicated
- The patient has the option to continue or terminate an affected pregnancy
  - We support patients and families in what ever decision is right for them

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Example 1 – sickle cell trait

- 19 year old female, African American G1P0

CBC W AUTOMATED DIFFERENTIAL

Status: Final result Visible to patient: Yes (kp.org) Next appt: 11/01/2017 at 10:40 AM in Obstetrics, Gynecology (LATONYA ROCHELLE BOTSHEKAN CNM,

	Ref Range & Units	9/22/17 1:37 PM	2/28/17 3:00 PM
WBC'S AUTO	4.0 - 11.0 x1000/mcL	6.1	5.9
RBC, AUTO	4.20 - 5.40 Mill/mcL	4.45	4.56
HGB	12.0 - 16.0 g/dL	12.0	12.6
HCT, AUTO	37.0 - 47.0 %	36.2 ↓	37.6
MCV	81.0 - 99.0 fL	81.2	82.4
MCH	27.0 - 35.0 pg/cell	27.0	27.7
MCHC	32.0 - 37.0 g/dL	33.2	33.6
RDW, BLOOD	11.5 - 14.5 %	16.7 ↑	12.9
PLATELETS, AUTOMATED COUNT	130 - 400 x1000/mcL	220	163
MPV			

HEMOGLOBIN VARIANT CONFIRMATION, ELECTROPHORESIS

Status: Final result Visible to patient: No (Not Released) Next appt: 11/01/2017 at 10:40 AM in Obstetrics, Gynecology (LATONYA ROCHELLE BOTSHEKAN CNM, C.N.M.)

	Ref Range & Units	9/22/17 1:37 PM
HGB S %, BLOOD, ELECTROPHORESIS	%	33.8
HEMOGLOBIN PHENOTYPE, BLOOD	AA	AS ↑
HGB PHENOTYPE INTERPRETATION, BLOOD, ELECTROPHORESIS		Heterozygous Hb AS ...

Heterozygous Hb AS (Hb AS Trait). This is a clinically benign condition which does not produce anemia. Hematology consultation is not necessary, but reproductive counseling may be advisable.

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Example 1 – FOB

- 21 year old male, African American

CBC NO DIFF
Status: Final result Visible to patient: kp.org Next appt: None Dx: SUPERVISION NORMAL FIRST PREGNANCY, F...

Newer results are available. Click to view them now.

	Ref Range	1/13/15 10:03 AM
WBC'S, AUTO	4.0 - 11.0 x1000/mcL	4.4
RBC, AUTO	4.20 - 5.40 Mill/mcL	4.47
HGB	12.0 - 16.0 g/dL	13.2
HCT, AUTO	37.0 - 47.0 %	39.0
MCV	81.0 - 99.0 fL	87.2
MCH	27.0 - 35.0 pg/cell	29.5
MCHC	32.0 - 37.0 g/dL	33.8
RDW, BLOOD	11.5 - 14.5 %	13.1
PLATELETS, AUTOMATED COUNT	130 - 400 x1000/mcL	157

HEMOGLOBIN VARIANT CONFIRMATION, ELECTROPHORESIS
Status: Final result Visible to patient: No (Not Released) Next appt: 11/01/2017 at 10:40 AM in Obstetrics, Gynecology (LATONYA ROCHELLE BOTSHEKAN CNM, C.N.M.)

	Ref Range & Units	9/22/17 1:37 PM
HGB S %, BLOOD, ELECTROPHORESIS	%	33.8
HEMOGLOBIN PHENOTYPE, BLOOD	AA	AS ↑
HGB PHENOTYPE INTERPRETATION, BLOOD, ELECTROPHORESIS		Heterozygous Hb AS ...

Heterozygous Hb AS (Hb AS Trait). This is a clinically benign condition which does not produce anemia. Hematology consultation is not necessary, but reproductive counseling may be advisable.

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Punnett Square

- Mother and father have Hb AS (sickle cell trait)

- ¼ chance for AA usual
- ½ chance for AS trait
- ¼ chance for sickle cell disease

Father \ Mother	A	S
A	AA	AS
S	AS	SS

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CALIFORNIA DEPARTMENT OF PUBLIC HEALTH
NEWBORN SCREENING PROGRAM
850 MARINA BAY PARKWAY, ROOM F175
RICHMOND, CA 94604

NEWBORN SCREENING RESULTS - INITIAL

[redacted] BABY
Gender: Male

HEMOGLOBIN INFORMATION
Date Time
Baby's Birth:
Specimen Collection:
Age at Collection:
Birth Weight:
Ethnicity: Black
Specimen Collection Site: KAISER-WEST LOS ANGEL
Feeding Type: Only Human Milk
Newborn on TPN/Hyperal or Amino Acids: No

[redacted] MOTHER

2 [redacted] SNGLP
T1 P1 06 -(300) 051101637360
KAISER-WEST LOS ANGELES
HOSP MED RECS/Andrea
6041 CADILLAC AVE
LOS ANGELES CA 90034-1702

Newborn's Physician: BENJAMIN FASS, MD

Testing Laboratory:
KAISER SOUTH BIOCHEMICAL GENETICS
4580 ELECTRONICS PL, LOS ANGELES, CA 90039-1008

Directors:
Alan H. Wu, Ph.D., DABCC, Genetic Disease Laboratory, (510) 231-1748

TEST - Individual analytes listed on reverse side	INTERPRETATION
Cystic Fibrosis (CF)	Negative
Biotinidase Deficiency	Negative
Galactosemia	Negative
Primary Congenital Hypothyroidism	Negative
Congenital Adrenal Hyperplasia (due to 21-Hydroxylase Deficiency)	Negative
MS/MS Acylcarnitine Panel	Negative
MS/MS Amino Acid Panel (Excluding PKU)	Negative
Severe Combined Immunodeficiency (SCID)	Negative
Hemoglobinopathies	**INTERPRETATION**
Hb Pattern: FAS	Consistent with sickle cell trait or a rare hemoglobin trait. Traits are clinically benign carrier conditions.

Follow-up:
Hb: Family testing is recommended because it may identify the possibility of sickle cell disease in future pregnancies. Hb trait counseling and family testing can be obtained by calling toll-free 1-800-734-5155.

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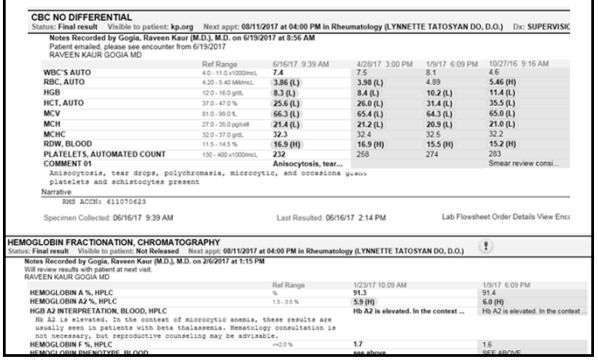
Example 2 – beta thalassemia trait

- 31 year old female patient, Asian East Indian



CBC NO DIFFERENTIAL

Status: **Final result** Visible to patient: **kp.org** Next appt: **08/11/2017 at 04:00 PM in Rheumatology (LYNNETTE TATOSYAN DO, D.O.)** Dx: **SUPERVISIC**

Notes Recorded by Gogia, Raveen Kaur (M.D.), M.D. on 6/19/2017 at 8:56 AM
Patient emailed, please see encounter from 6/19/2017
RAVEEN KAUR GOGIA MD

	Ref Range	6/16/17 9:39 AM	4/28/17 3:00 PM	1/9/17 6:09 PM	10/27/16 9:16 AM
WBC'S AUTO	4.0 - 11.0 x1000/mcL	7.4	7.5	8.1	4.6
RBC, AUTO	4.20 - 5.40 Mill/mcL	3.86 (L)	3.98 (L)	4.89	5.46 (H)
HGB	12.0 - 16.0 g/dL	8.3 (L)	8.4 (L)	10.2 (L)	11.4 (L)
HCT, AUTO	37.0 - 47.0 %	25.6 (L)	26.0 (L)	31.4 (L)	35.5 (L)
MCV	81.0 - 99.0 fL	66.3 (L)	65.4 (L)	64.3 (L)	65.0 (L)
MCH	27.0 - 35.0 pg/cell	21.4 (L)	21.2 (L)	20.9 (L)	21.0 (L)
MCHC	32.0 - 37.0 g/dL	32.3	32.4	32.5	32.2
RDW, BLOOD	11.5 - 14.5 %	16.9 (H)	16.9 (H)	15.5 (H)	15.2 (H)
PLATELETS, AUTOMATED COUNT	130 - 400 x1000/mcL	232	258	274	283
COMMENT 01		Anisocytosis, tear...			Smear review consi...

Anisocytosis, tear drops, polychromasia, microcytic, and occasiona
platelets and schistocytes present

Narrative

RMS ACCN: 611070623

Specimen Collected: 06/16/17 9:39 AM Last Resulted: 06/16/17 2:14 PM Lab Flowsheet Order Details View Enc

HEMOGLOBIN FRACTIONATION, CHROMATOGRAPHY

Status: **Final result** Visible to patient: **Not Released** Next appt: **08/11/2017 at 04:00 PM in Rheumatology (LYNNETTE TATOSYAN DO, D.O.)**

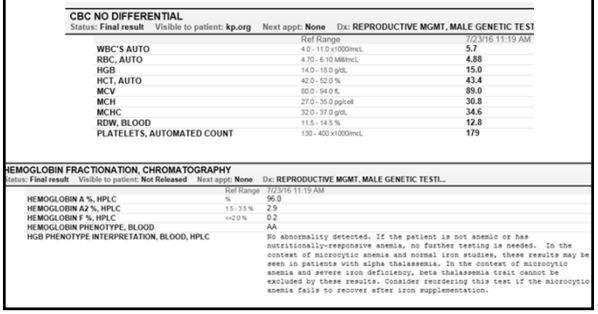
Notes Recorded by Gogia, Raveen Kaur (M.D.), M.D. on 2/6/2017 at 1:15 PM
Will review results with patient at next visit.
RAVEEN KAUR GOGIA MD

	Ref Range	1/23/17 10:09 AM	1/9/17 6:09 PM
HEMOGLOBIN A %, HPLC	%	91.3	91.4
HEMOGLOBIN A2 %, HPLC	1.5 - 3.5 %	5.9 (H)	6.0 (H)
HGB A2 INTERPRETATION, BLOOD, HPLC		Hb A2 is elevated. In the context ...	Hb A2 is elevated. In the context ...
HEMOGLOBIN F %, HPLC	<=2.0 %	1.7	1.6
HEMOGLOBIN PHENOTYPE, BLOOD		see above	SEE ABOVE

Hb A2 is elevated. In the context of microcytic anemia, these results are usually seen in patients with beta thalassemia. Hematology consultation is not necessary, but reproductive counseling may be advisable.

Example 2 - FOB

- 31 year old male partner, Chinese ancestry



CBC NO DIFFERENTIAL

Status: **Final result** Visible to patient: **kp.org** Next appt: **None** Dx: **REPRODUCTIVE MGMT, MALE GENETIC TEST**

	Ref Range	7/23/16 11:19 AM
WBC'S AUTO	4.0 - 11.0 x1000/mcL	5.7
RBC, AUTO	4.70 - 6.10 Mill/mcL	4.88
HGB	14.0 - 18.0 g/dL	15.0
HCT, AUTO	42.0 - 52.0 %	43.4
MCV	80.0 - 94.0 fL	89.0
MCH	27.0 - 35.0 pg/cell	30.8
MCHC	32.0 - 37.0 g/dL	34.6
RDW, BLOOD	11.5 - 14.5 %	12.8
PLATELETS, AUTOMATED COUNT	130 - 400 x1000/mcL	179

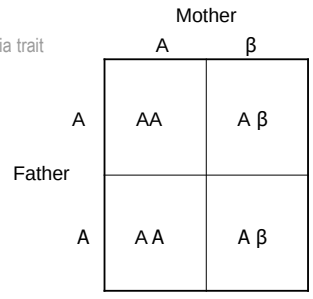
HEMOGLOBIN FRACTIONATION, CHROMATOGRAPHY

Status: **Final result** Visible to patient: **Not Released** Next appt: **None** Dx: **REPRODUCTIVE MGMT, MALE GENETIC TESTI...**

	Ref Range	7/23/16 11:19 AM
HEMOGLOBIN A %, HPLC	%	96.0
HEMOGLOBIN A2 %, HPLC	1.5 - 3.5 %	2.9
HEMOGLOBIN F %, HPLC	<=2.0 %	0.2
HEMOGLOBIN PHENOTYPE, BLOOD		AA
HGB PHENOTYPE INTERPRETATION, BLOOD, HPLC		No abnormality detected. If the patient is not anemic or has nutritionally-responsive anemia, no further testing is needed. In the context of microcytic anemia and normal iron studies, these results may be seen in patients with alpha thalassemia. In the context of microcytic anemia and severe iron deficiency, beta thalassemia trait cannot be excluded by these results. Consider reordering this test if the microcytic anemia fails to recover after iron supplementation.

Punnett Square

- Mother has beta thalassemia trait and father is normal



- 1/2 chance for AA usual
- 1/2 chance for beta thal trait

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Example 3 – beta thalassemia trait

- 31 year old female patient, Asian East Indian ancestry

CBC NO DIFFERENTIAL

Status: Final result Visible to patient: No (Not Released) Next appt: 10/19/2017 at 09:20 AM in Obstetrics, Gynecology (JENNIFER JEAN LEE MD, M.D.)

	Ref Range & Units	9/14/17 8:14 AM
WBC'S AUTO	4.0 - 11.0 x1000/mcL	5.4
RBC, AUTO	4.20 - 5.40 Mill/mcL	5.53
HGB	12.0 - 16.0 g/dL	10.9
HCT, AUTO	37.0 - 47.0 %	34.8
MCV	81.0 - 99.0 fL	63.0
MCH	27.0 - 35.0 pg/cell	19.7
MCHC	32.0 - 37.0 g/dL	31.2
RDW, BLOOD	11.5 - 14.5 %	15.5
PLATELETS, AUTOMATED COUNT	130 - 400 x1000/mcL	200

HEMOGLOBIN FRACTIONATION, CHROMATOGRAPHY

Status: Final result Visible to patient: No (Not Released) Next appt: 10/19/2017 at 09:20 AM in Obstetrics, Gynecology (JENNIFER JEAN LEE MD, M.D.) Dx: ANTENATAL SCREENING

	Ref Range & Units	9/14/17 8:14 AM
HEMOGLOBIN A %, HPLC	%	91.5
HEMOGLOBIN A2 %, HPLC	1.5 - 3.5 %	5.6
HGB A2 INTERPRETATION, BLOOD, HPLC		Hb A2 is elevated. In the context of microcytic anemia, these results are usually seen in patients with beta thalassemia. Hematology consultation is not necessary, but reproductive counseling may be advisable.
HEMOGLOBIN F %, HPLC	<=2.0 %	1.7
HEMOGLOBIN PHENOTYPE, BLOOD		see above

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Example 3 - FOB

- 34 year old male partner, Asian East Indian ancestry

CBC NO DIFFERENTIAL

Status: Final result Visible to patient: kp.org Next appt: None Dx: GENETIC COUNSELING

	Ref Range	2/10/17 4:15 PM	Flag
WBC'S AUTO	4.0 - 11.0 x1000/mcL	7.5	
RBC, AUTO	4.70 - 6.10 Mill/mcL	6.63	H
HGB	14.0 - 18.0 g/dL	13.6	L
HCT, AUTO	42.0 - 52.0 %	42.1	
MCV	80.0 - 94.0 fL	63.5	L
MCH	27.0 - 35.0 pg/cell	20.5	L
MCHC	32.0 - 37.0 g/dL	32.3	
RDW, BLOOD	11.5 - 14.5 %	14.8	H
PLATELETS, AUTOMATED COUNT	130 - 400 x1000/mcL	201	

HEMOGLOBIN FRACTIONATION, CHROMATOGRAPHY

Status: Final result Visible to patient: Not Released Next appt: None Dx: GENETIC COUNSELING

	Ref Range	2/10/17 4:15 PM	Flag
HEMOGLOBIN A %, HPLC	%	93.4	
HEMOGLOBIN A2 %, HPLC	1.5 - 3.5 %	4.8	H
HGB A2 INTERPRETATION, BLOOD, HPLC		Hb A2 is elevated. In the context of microcytic anemia, these results are usually seen in patients with beta thalassemia. Hematology consultation is not necessary, but reproductive counseling may be advisable.	
HEMOGLOBIN F %, HPLC	<=2.0 %	0.4	
HEMOGLOBIN PHENOTYPE, BLOOD		SEE ABOVE	

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Punnett Square

- Mother and father both have beta thalassemia trait

Father \ Mother	A	β
A	AA	A β
β	A β	β β

- ¼ chance for AA usual
- ½ chance for beta thal trait
- ¼ chance for beta thal disease

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TPMG REGIONAL GENETICS LABORATORY
MOLECULAR GENETIC REPORT

Report# 389546
Page: 1

Name : [redacted] (Fetus Of)
MR# :
DOB :
Sex :U
Lab# :
Ped# :
Date Received :03/02/17
Date of Report :03/10/17
Region :So Cal Kaiser

Req Facility :Baldwin Park
Req Provider :Cristiane Guberman, MD
Gen Center :West Los Angeles
Gen Counselor :Emily S Parkhurst, MS, LCGC
Med Geneticist:
Date Obtained :02/28/17
Specimen :CVS-direct
Outside Ref# :

Indication:
Prenatal diagnosis of beta thalassemia. The mother of the fetus is a carrier for the 619bp deletion and the father is a carrier for the c.92+1G>T variant within the HBB gene.

REPORT

Test: DNA analysis of the Beta globin gene.

Result:
DNA sequence analysis revealed the presence of a known pathogenic variant, c.92+1G>T, within the HBB gene. The 619bp deletion was not detected in this fetal sample.

INTERPRETATION:
This fetus is a carrier for the c.92+1G>T (beta 0) variant within the HBB gene. The study for maternal cell contamination was not performed based on this result.

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Example 4 – iron deficiency

- 30 year old Mexican American G3P2

CBC NO DIFFERENTIAL

Status: Final result Visible to patient: Yes (kp.org) Next appt: 10/14/2017 at 01:00 PM in Health Education (MEDYLINE BULANTE RN, R.N.) Dx: ANTI

Newer results are available. C

	Ref Range & Units	4/4/17 1:52 PM
WBC'S AUTO	4.0 - 11.0 x1000/mcL	6.2
RBC, AUTO	4.20 - 5.40 Mill/mcL	4.53
HGB	12.0 - 16.0 g/dL	11.0
HCT, AUTO	37.0 - 47.0 %	33.9
MCV	81.0 - 99.0 fL	74.9
MCH	27.0 - 35.0 pg/cell	24.2
MCHC	32.0 - 37.0 g/dL	32.4
RDW, BLOOD	11.5 - 14.5 %	16.3
PLATELETS, AUTOMATED COUNT	130 - 400 x1000/mcL	238

HEMOGLOBIN FRACTIONATION, CHROMATOGRAPHY

Status: Final result Visible to patient: No (Not Released) Next appt: 10/14/2017 at 01:00 PM in Health Education (MEDYLINE BULANTE RN, R.N.) Dx: ANTENATAL SCREENING

	Ref Range & Units	4/4/17 1:52 PM
HEMOGLOBIN A %, HPLC	%	95.8
HEMOGLOBIN A2 %, HPLC	1.5 - 3.5 %	2.8
HEMOGLOBIN F %, HPLC	<=2.0 %	0.2
HEMOGLOBIN PHENOTYPE, BLOOD		AA
HGB PHENOTYPE INTERPRETATION, BLOOD, HPLC		No abnormality detected.

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Example 4 – iron deficiency

- 30 year old Mexican American G3P2

IRON AND TIBC

Status: Final result Visible to patient: Yes (kp.org) Next appt: 10/14/2017 at 01:00 PM in Health Education (MEDYLINE BULANTE RN, R.N.) Dx: ANEMIA IN PREG

	Ref Range & Units	6/1/17 10:48 AM
IRON	37 - 145 mcg/dL	44
TOTAL IRON BINDING CAPACITY	250 - 450 mcg/dL	436
IRON SAT	20 - 50 %	10

Narrative
RMS ACCN: 610029443

Specimen Collected: 06/01/17 10:48 AM Last Resulted: 06/01/17 5:16 PM

- FOB is also Mexican-American. Since the couple is not of a high risk ancestry, the risk is NOT at increased for a hemoglobinopathy.
- Results consistent with iron deficiency

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Example 5 – alpha thalassemia trait

- 24 year old African American G2P1

CBC NO DIFFERENTIAL

Status: Final result Visible to patient: Yes (kp.org) Next appt: None Dx: ROUTINE ADULT HEALTH CHECK UP EXAM

Notes Recorded by Tuason, Angelica S (R.N.), R.N. on 8/8/2017 at 10:14 AM
Letter sent

******

Notes Recorded by Zheng, Xiaona (M.D.), M.D. on 8/7/2017 at 6:48 PM
Call or letter - Blood test is normal, no more anemia

	Ref Range & Units	8/7/17 9:59 AM	1/13/16 12:30 PM
WBC'S AUTO	4.0 - 11.0 x1000/mcL	5.5	7.8
RBC, AUTO	4.20 - 5.40 Mill/mcL	5.65 ^	5.38
HGB	12.0 - 16.0 g/dL	12.5	11.4 v
HCT, AUTO	37.0 - 47.0 %	39.0	36.5 v
MCV	81.0 - 99.0 fL	69.1 v	67.9 v
MCH	27.0 - 35.0 pg/cell	22.2 v	21.2 v
MCHC	32.0 - 37.0 g/dL	32.1	31.2 v
RDW, BLOOD	11.5 - 14.5 %	14.7 ^	18.9 ^
PLATELETS, AUTOMATED COUNT	130 - 400 x1000/mcL	222	293

HEMOGLOBIN FRACTIONATION, CHROMATOGRAPHY

Status: Final result Visible to patient: No (Not Released) Next appt: None Dx: THALASSEMIA; SCREENING FOR GENETIC DI...

	Ref Range & Units	9/11/17 12:28 PM
HEMOGLOBIN A %, HPLC	%	95.7
HEMOGLOBIN A2 %, HPLC	1.5 - 3.5 %	2.7
HEMOGLOBIN F %, HPLC	<=2.0 %	0.4
HEMOGLOBIN PHENOTYPE, BLOOD		AA
HGB PHENOTYPE INTERPRETATION, BLOOD, HPLC		No abnormality detected.

Example 5 – alpha thalassemia trait

- 24 year old African American G2P1

IRON AND TIBC

Status: Final result Visible to patient: Yes (kp.org) Next appt: None Dx: THALASSEMIA

	Ref Range & Units	9/11/17 12:28 PM
IRON	37 - 145 mcg/dL	177 ^
TOTAL IRON BINDING CAPACITY	250 - 450 mcg/dL	361
IRON SAT	20 - 50 %	49

FERRITIN

Status: Final result Visible to patient: Yes (kp.org) Next appt: None Dx: THALASSEMIA

	Ref Range & Units	9/11/17
FERRITIN	13 - 126 ng/mL	34

Comments: Iron therapy of anemic chronic kidney disease and dialysis patients may be adjusted to produce serum ferritin levels between 100 and 1200 ng/ml.

- FOB is also African-American. Since the couple is not of a high risk ancestry, the risk is NOT increased for hemoglobinopathy.
- Patient told to stop iron supplements

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Example 6 – Hb C trait

- 33 year old G3P2 African-American woman

HEMOGLOBIN VARIANT CONFIRMATION, ELECTROPHORESIS

Status: Final result Visible to patient: No (Not Released) Next appt: None

	Ref Range & Units	12/21/16 11:39 AM
HEMOGLOBIN A,BLD,QN, ELECTROPHORESIS	%	59.6
HGB C %, BLOOD, ELECTROPHORESIS	%	36.8
HEMOGLOBIN PHENOTYPE, BLOOD	AA	AC !
HGB PHENOTYPE INTERPRETATION, BLOOD, ELECTROPHORESIS		Heterozygous Hb C (...

Heterozygous Hb C (Hb C Trait). This is a clinically benign condition which does not produce anemia. Hematology consultation is not necessary, but reproductive counseling may be advisable. Anemia, preponderance of Hb C and Hb F elevations may indicate the presence of an underlying thalassemia.

Narrative

RMS ACCN: 598936625

Specimen Collected: 12/21/16 11:39 AM Last Resulted: 12/23/16 2:31 PM

x=Reference range differs from displayed range

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Example 6 – FOB

- 36 year old African-American man

HEMOGLOBIN FRACTIONATION, CHROMATOGRAPHY

Status: Final result Visible to patient: No (Not Released) Next appt: None Dx: REPRODUCTIVE MGMT, MALE GENETIC TESTI...

	Ref Range & Units	8/30/17 2:04 PM
HEMOGLOBIN A %, HPLC	%	95.9
HEMOGLOBIN A2 %, HPLC	1.5 - 3.5 %	2.7
HEMOGLOBIN F %, HPLC	<=2.0 %	0.2
HEMOGLOBIN PHENOTYPE, BLOOD		AA
HGB PHENOTYPE INTERPRETATION, BLOOD, HPLC		No abnormality detected.

CBC NO DIFFERENTIAL

Status: Final result Visible to patient: No (Not Released) Next appt: None Dx: REPRODUCTIVE MGMT, MALE GENETIC TESTI...

	Ref Range & Units	8/30/17 2:04 PM
WBC'S AUTO	4.0 - 11.0 x1000/mcL	4.2
RBC, AUTO	4.70 - 6.10 Mill/mcL	5.75
HGB	14.0 - 18.0 g/dL	13.4
HCT, AUTO	42.0 - 52.0 %	41.7
MCV	80.0 - 94.0 fL	72.5
MCH	27.0 - 35.0 pg/cell	23.3
MCHC	32.0 - 37.0 g/dL	32.1
RDW, BLOOD	11.5 - 14.5 %	13.9
PLATELETS, AUTOMATED COUNT	130 - 400 x1000/mcL	195

- He may be a carrier of alpha thalassemia trait
- Couple not at risk to have a baby with a hemoglobinpathy

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Punnett Square

- Mother has Hb C trait and father may have alpha thalassemia trait
- Since the couple is of African ancestry, the risk is not increased for alpha thalassemia major

•½ chance for Hb AC
•½ chance for Hb AA

•Because Hb C is a beta globin mutation and alpha thalassemia is due to an alpha globin mutation, these do not combine to cause disease

		Mother A	Mother C
Father	A	AA	AC
	A	A A	AC

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Example 7 – HPFH

- 31 year old G2P1 European-American woman

HEMOGLOBIN FRACTIONATION, CHROMATOGRAPHY

Status: Final result Visible to patient: No (Not Released) Next appt: None Dx: PRENATAL INTAKE INTERVIEW

	Ref Range & Units	8/25/17 3:17 PM
HEMOGLOBIN A %, HPLC	%	72.6
HEMOGLOBIN A2 %, HPLC	1.5 - 3.5 %	2.2
HEMOGLOBIN F %, HPLC	<=2.0 %	25.5
HGB F INTERPRETATION, BLOOD, HPLC		Elevated Hb F. This...
HEMOGLOBIN PHENOTYPE, BLOOD		SEE ABOVE

Elevated Hb F. This pattern can occur in the presence of hereditary persistence of fetal hemoglobin (HPFH) or beta-thalassemia. Isolated HbF elevation can also occur with pregnancy, myeloproliferative disorders, and marrow failure. Up to 10% Hb F can be found in normal patients 1-2 years of age, but Hb F rarely exceeds 2% thereafter.

CBC NO DIFFERENTIAL

Status: Final result Visible to patient: Yes (kp.org) Next appt: None Dx: SUPERVISION NORMAL PREGNANCY, THIRD T...

	Ref Range & Units	4/21/16 11:27
WBC'S AUTO	4.0 - 11.0 x1000/mcL	9.9
RBC, AUTO	4.20 - 5.40 Mill/mcL	4.24
HGB	12.0 - 16.0 g/dL	11.5
HCT, AUTO	37.0 - 47.0 %	35.0
MCV	81.0 - 99.0 fL	82.4
MCH	27.0 - 35.0 pg/cell	27.2
MCHC	32.0 - 37.0 g/dL	33.0
RDW, BLOOD	11.5 - 14.5 %	14.0
PLATELETS, AUTOMATED COUNT	130 - 400 x1000/mcL	281
MPV		

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Refer to Genetics

When in doubt, send a Dr. Advice to genetics.

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