

# Sickle Cell Anemia

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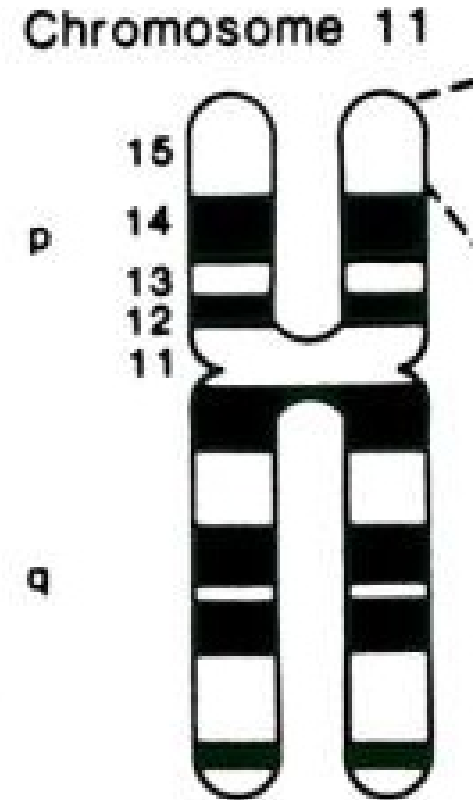
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# Sickle Cell Disease - Summary

- Autosomal Recessive Disease
- Most common blood disease in USA
  - Affects 72000 Americans
  - 1 in 500 African Americans
- Chromosome 11p15.5
  - Affect Beta Hemoglobin
- Missense Glu6Val mutation
  - Hydrophobic bonds
- Common in malaria hot spots
  - Africa, Mediterranean, Caribbean, and descendants



# Symptoms

- Pleiotropic Gene
  - RBC sickle- low pH or O<sub>2</sub>
- Homozygous Recessive
  - Blood Cell Destruction
  - Physical Weakness
  - Anemia
  - Circulatory disturbances
  - Impaired mental function
  - Organ Damage
  - Stroke
- Heterozygous
  - Sickling under low pH or O<sub>2</sub>
  - Fainting
  - Gross Hematuria
  - Leg Compartment Syndrome



# Traditional Diagnostic Exams

## ○ High-performance liquid chromatography (HPLC)

- Separates protein difficult to separate
- Easily see Hb S

## ○ Isoelectric focusing (IEF)

- Separated by pH at 0 charge

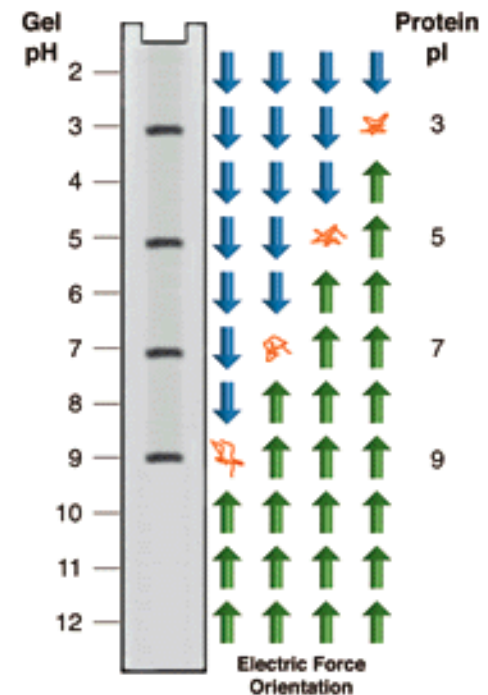
## ○ Cellulose acetate and citrate agar electrophoresis

- Useful for quick screenings

## ○ Peripheral blood smear

- Sickled cells and RBC easily seen

## ○ Focus of these treatments is to isolate mutant hemoglobin from bloodstream



# Traditional Treatments

## ○ Pain Treatments

- Warmth
- Massage
- Distraction
- Acupuncture
- Biofeedback
- Hypnosis

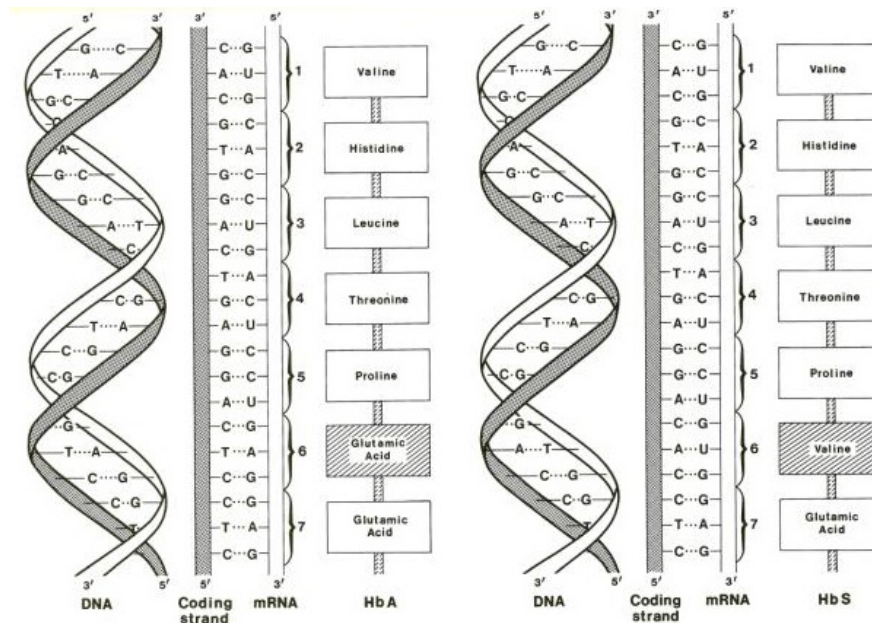
## ○ Supportive Treatments

- Hydration (via Intravenous Fluids)
- Anti- Inflammatory Agents
- Pain medication



# Novel Diagnostics

- Prenatal Screenings
- Targeted Mutation Analysis
  - Tests for Glu6Val Mutation
  - Can also test for other sickle cell diseases
- Sequence Analysis
  - Determine DNA Sequence
  - Used if mutation analysis fails



# Novel Treatments

- Prevention Treatments
- Primary Treatments
  - Hydroxeurea- Produces fetal Hb
  - Chronic Red Blood Cell Transfusion
  - Stem Cell Transplantation
- Therapies under Investigation
  - Gene Therapy
  - Gene Therapy using IPS



# Sources

- <http://www.ncbi.nlm.nih.gov/books/NBK1377/>
- <http://www.ncbi.nlm.nih.gov/books/NBK22238/>
- “Sickle Cell 2011 for Posting”- William Durham
- <http://biochem118.stanford.edu/Projects/2011%20Winter/Gabe-2011>

