Thalassemia:
Overview
Current Challenges and Opportunities

Sujit Sheth, M.D.

Weill Cornell Medicine

New York Comprehensive Thalassemia Center

NewYork-Presbyterian
Komansky Children's Hospital

SPEAKER DISCLOSURE

- Consultant
  - Celgene / Bristol Myers Squibb / Acceleron
  - Agios
  - Forma/ Novonordisk
- Steering committee
  - CRISPR/ Vertex CTX001
- Research funding
  - Agios
  - Bristol Myers Squib
  - Forma/Novonordisk
  - Regeneron

OUTLINE

- What is thalassemia?
- Current paradigm of diagnosis and treatment in the US
- Challenges in provision of comprehensive care
- Integration of novel therapies
- Opportunities to improve delivery of care

HEMOGLOBIN DISORDERS

Qualitative Hemoglobinopathies

Globin gene mutations that result in structural abnormalities of the globin chain:

Hb S, Hb C, Hb E and other Hb variants

Quantitative Hemoglobinopathies (disorders of ineffective erythropoiesis)

Globin gene mutations that result in decreased production of globin chains:

Thalassemias (Alpha, Beta, Gamma, Delta)

GENOTYPE-PHENOTYPE

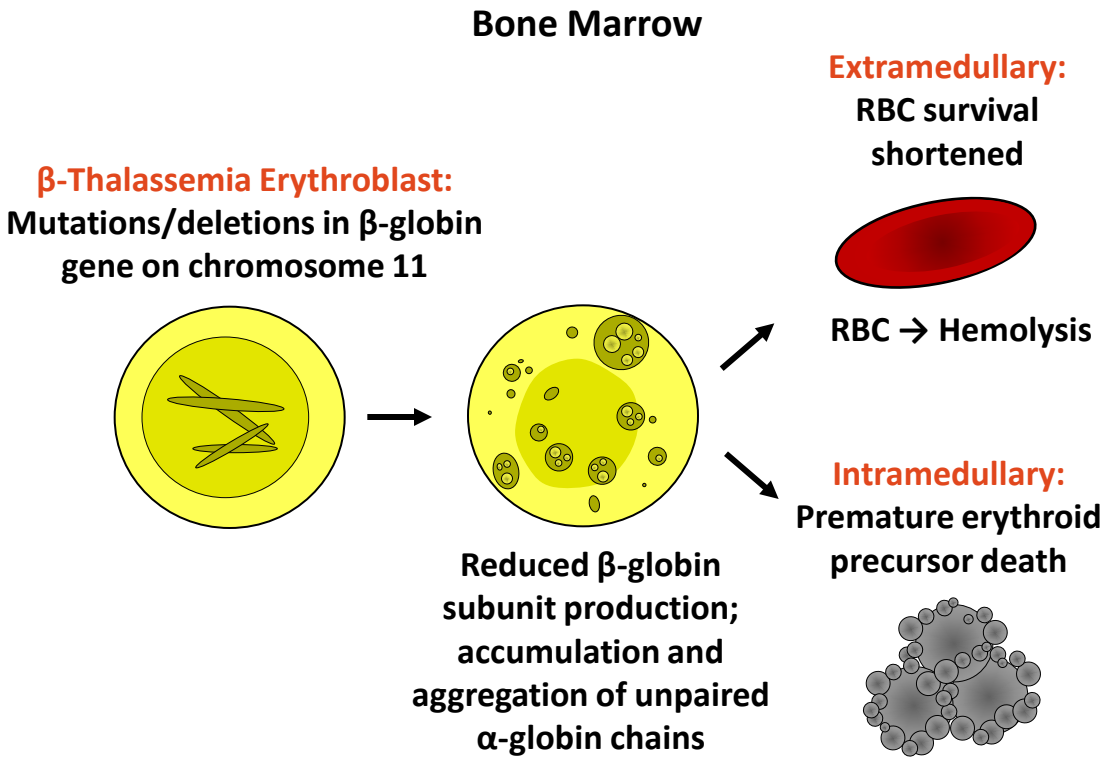
Mild	Non Transfusion dependent	Transfusion dependent
Anemia ranging from very mild to low end of normal	Intermediate severity Moderate anemia	Severe anemia
α-thalassemia trait/silent carrier	α-thalassemia intermedia-Hb H	α-thalassemia major/Hb Barts
β-thalassemia minor/trait	β-thalassemia intermedia	β-thalassemia major
	Dominant β-thalassemia	Severe Hb E β-thalassemia
	Hb H Constant Spring	Severe Hb H Constant Spring
	Hemoglobin E β-thalassemia	

NOT SO BENIGN

HEMOGLOBIN ABNORMALITY

- Imbalance between normal ratio of α : β chain production
- Insufficient upregulation of complementary genes
- Precipitation of tetramers, formation of hemichromes
- Cell disruption - apoptosis
- Ineffective erythropoiesis

FEATURES OF INEFFECTIVE ERYTHROPOIESIS

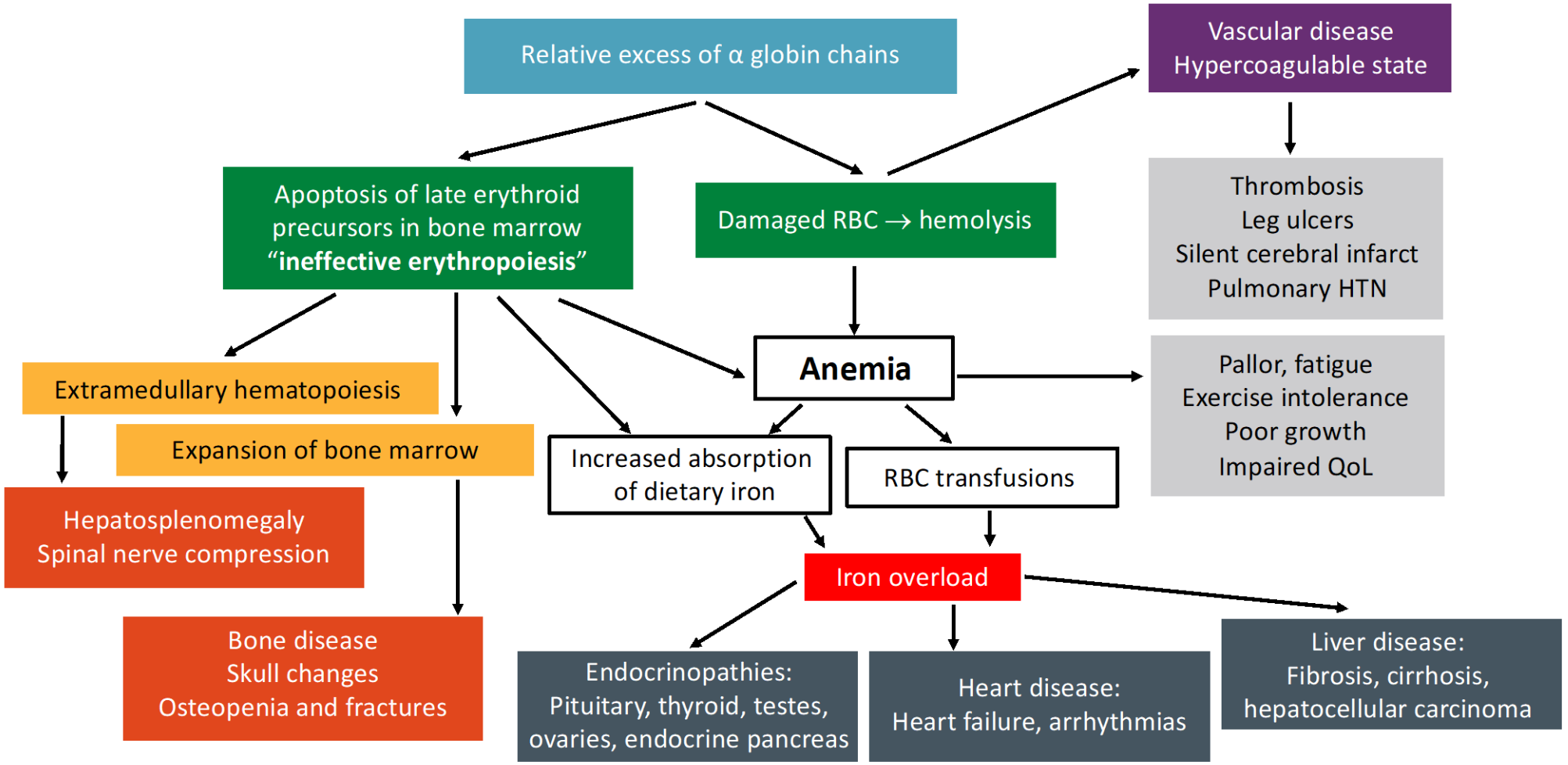


- Impaired erythroid precursor maturation
- α and β chain imbalance
- Formation of toxic hemichromes from precipitation of unpaired α-globin chains
- Apoptosis of erythroid precursors
- Reduced RBC survival
- Anemia
- Increased erythropoietic drive
- Extramedullary hematopoiesis
- Dysregulated iron metabolism

SPECTRUM OF DISEASE

Syndrome	Genotype	Hematology	Disease Severity
Thalassemia major	β^0/β^0	▪ Complete absence of Hb A ▪ Severe anemia requiring transfusions from infancy	▪ **TD** ▪ Lifelong supportive care required
Thalassemia intermedia	β^+/β^+ or β^0/β^+	▪ Diminished production of Hb A ▪ Mild to moderate anemia	▪ **NTD** ▪ May need occasional transfusions or may become **TD** ▪ Significant variability in disease severity
Thalassemia minor	β^+/β or β^0/β	▪ Mild or no anemia	▪ **NTD** ▪ May be asymptomatic

PATHOLOGY AND CLINICAL FEATURES



DIAGNOSIS

- History - family history, ethnicity
- Antenatal/ couples screening
- Newborn screening
- Clinical syndrome - anemia, hepatosplenomegaly, facies, skeletal abnormalities
- CBC - anemia with low MCV, low MCH, smear
- Hemoglobin electrophoresis
- Genetic testing

CLINICAL MANIFESTATIONS

- **Anemia:** Impaired function, impaired growth, impaired QoL
- **Iron overload:** Increased GI absorption, transfused iron
- **Cardiac disease:** Anemia, iron deposition
- **Endocrinopathy:** Pituitary, thyroid, endocrine pancreas, gonads
- **Bone disease:** Erythroid hyperplasia, endocrinopathy
- **Gallbladder disease:** Increased red cell turnover
- **Extramedullary hematopoiesis:** Hepatosplenomegaly, spinal nodules
- **Vascular disease:** Leg ulcers, pulmonary hypertension, stroke

Typical Course in Transfusion Dependent Thalassemia

FIRST DECADE

Initiate regular monthly RBC transfusions at young age

Start iron chelation

Annual liver MRI from age 2 yrs and chelator associated monitoring

SECOND DECADE

Increased transfusion - 2-3 units every 2-5 weeks

Continue chelation – reinforce compliance

Annual liver and cardiac MRI after age 10 years
Chelator associated monitoring

ADULTHOOD

Ongoing regular transfusions & chelation

Annual MRI: Liver, Cardiac
Chelator associated monitoring
Bone density
Endocrine assessments

Iron Overload Complications

Delayed Growth	Delayed puberty Diabetes Other endocrinopathies Heart failure	Secondary amenorrhea Infertility Osteoporosis and fractures Liver disease

COURSE - NTDT

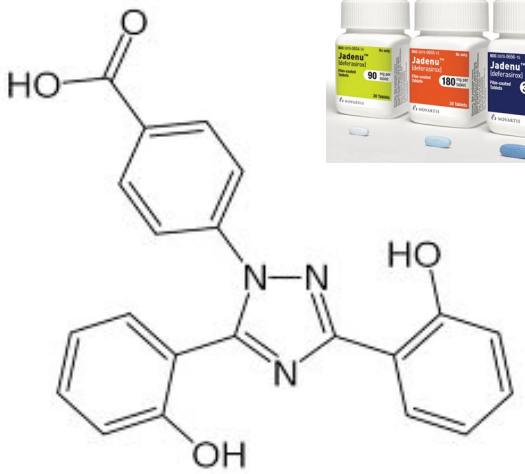
- Supportive care: Bone health, vitamin D and folic acid supplementation, thromboprophylaxis
- Splenectomy: If severe anemia and splenomegaly
- Transfusions (periodic): Leg ulcers, splenomegaly, pregnancy, surgery
- Iron chelation
- Induction of Hb F: Hydroxyurea, 5’azacytidine, decitabine, butyrate
- Other: Luspatercept, ruxolitinib
- ?Stem cell transplantation, ?gene therapy

MANAGEMENT ISSUES

- Transfusions, splenectomy
- Consideration for luspatercept
- Transfusion complications
- Complications of Iron overload
- Chelation - Side effects and monitoring
- Organ dysfunction
- Consideration for stem cell transplantation
- Newer therapies

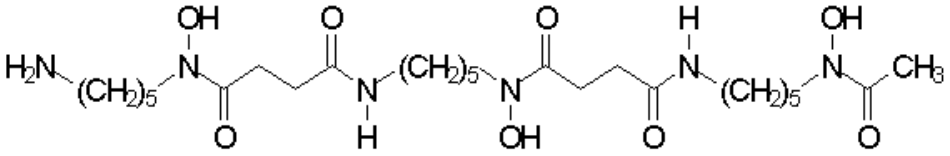
IRON CHELATION

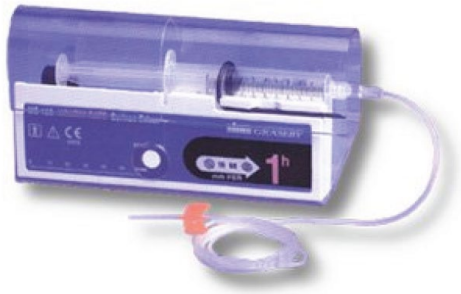
DEFERASIROX
(Exjade®, Jadenu®)



DEFERIPRONE
(Ferriprox®)

DEFEROXAMINE
(Desferal®)





GENE THERAPY APPROACHES

- Globin gene addition
  - Functional β-globin gene
    - » LentiGlobin BB305
    - » GLOBE lentiviral vector
  - Functional γ-globin gene
- Gene editing
  - Reverse γ-globin repression
    - » Targeting BCL11A
      - CRISPR/CAS9 gene-editing - CTX001
    - » Targeting γ-globin gene promoters
      - CRISPR-Cas12a editing – EDIT-301
  - Correction of the β-globin mutation

APPROVED THERAPIES
-Zynteglo®
-Casgevy®

GENE THERAPY - Conclusions

- β-globin gene addition and editing trials have shown positive results
  - High rate of transfusion independence across all genotypes
  - Long-term data with high VCN / high degree of editing
  - Hemoglobin levels are in normal or near normal ranges
  - Response is durable
  - Improvements in overall HRQoL
- No deaths to date, generally well tolerated in pediatric and adult patients
- Toxicity mostly related to myeloablative conditioning
- Risk for insertional mutagenesis (addition) or off-target effects (editing)need careful follow-up
- Careful patient selection necessary, with attention to individual needs, expectations and eligibility

QUALITY OF LIFE

- Issues related to:
  - Symptoms – fatigue
  - Physical appearance (NTDT)
  - Frequent visits to the hospital
  - Need for chelation compliance
  - Pain - Bone disease, extramedullary hematopoiesis (NTDT)
  - Endocrine – growth, development, fertility
  - Financial issues
- Psychosocial issues
  - Chronic illness
  - Reduced life expectancy with complications

Treatments

- Non-curative
  - Supportive care
  - Transfusions – regular or episodic
  - Iron chelation
  - Hydroxyurea, butyrate
  - Luspatercept
  - Thalidomide
- Curative
  - HSCT
    - » MRD
    - » MUD, Haplo

Progress

- Improved safety of blood supply
- Reduced incidence of alloimmunization
- Oral iron chelation
- Improved monitoring of iron overload to enable individualized tailoring of treatment regimen
- Improved outcomes for stem cell transplantation
- Overall increased survival

Unmet Needs

- Access to comprehensive care
- Access to novel disease modifying treatments
- Means of ameliorating ineffective erythropoiesis
- Prevent iron overload and its complications
- Reduce bone disease
- Improve quality of life
- **Curative options for all**

HEALTH ECONOMICS

- Annual healthcare costs for TDT patient - ~$130K in 2016
- Improved TDT survival and complication free survival on conventional therapies – even without luspatercept
- Additional cost of ~$60-80K for luspatercept
- Cost of Zynteglo® - $3.1 million
- Cost of Casgevy® - $2.2 million
- Annual cost of Pyrukynd® for PK deficiency - $300-330K

Future Directions

- Pyruvate kinase activators in clinical practice
- Other novel approaches – HbF induction, modification of the iron metabolism pathway
- Non-chemotherapy conditioning for HSCT and gene therapy
- Improved fertility preservation
- In-vivo gene therapy – editing/insertion - nanoparticles

CHALLENGES

- Identification of patients
  - Couples testing - ?type of testing
  - Prenatal diagnosis
  - Newborn screening – variable from state to state
  - Immigrants and refugees – often in areas without thalassemia resources
- Accurate and timely diagnosis
- Patients not well versed, cannot advocate for themselves
- Access to comprehensive care – multispecialty medical home
- Guidelines exist, but awareness is inadequate

ADVANCES

- HRSA and CDC efforts
  - Identification of patients
  - Informing hematologists
  - Creation of patient facing materials
- Cooley's Anemia Foundation efforts
  - Checklists
  - Patient and Family day
  - Scientific efforts – research grants, symposium every 5 years

OPPORTUNITIES

- Uniform Newborn Screening practices – genetic platform
- Identifying at risk populations – needs more attention given increases in immigration and refugee resettlement from Asia
- Establishing a comprehensive care guideline including monitoring for NTDT patients as well
- Enhancing patient and provider information levels
  - ?American Society of Hematology focus, like with SCD
  - Using the Thalassemia International Federation (TIF) model
- Creating materials to assist decision making around novel therapies, especially gene therapy

Areas of Opportunity: Optimizing Care and Management of Individuals, Families, and Caregivers Living with Thalassemia

Craig Butler

National Executive Director

Cooley's Anemia Foundation

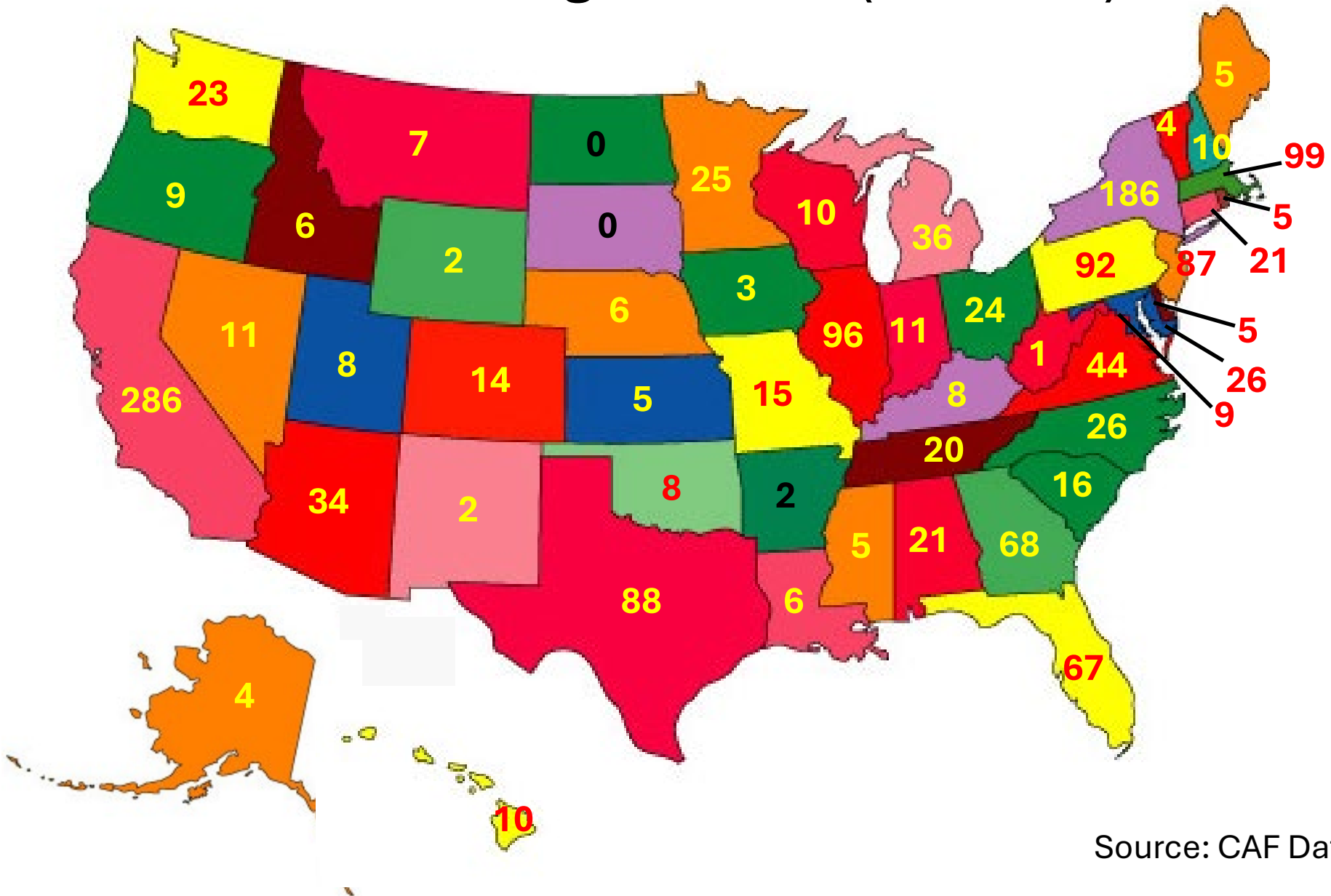
Cooley's Anemia Foundation
70 Years of Leading the Fight Against Thalassemia

Cooley's Anemia Foundation

- Founded 1954
- Serves Individuals with Severe Forms of Thalassemia in the U.S.
  - Not Trait
- 1555 in Database
  - About Half of US Patients?
- Medical Research
- Communications
  - Outreach to Communities
- Patient Services
  - Annual Conference
  - Support Programs
  - Informational Resources

Cooley's Anemia Foundation
70 Years of Leading the Fight Against Thalassemia

Thalassemia Patient Geographical Distribution Throughout U.S. (n=1,555)



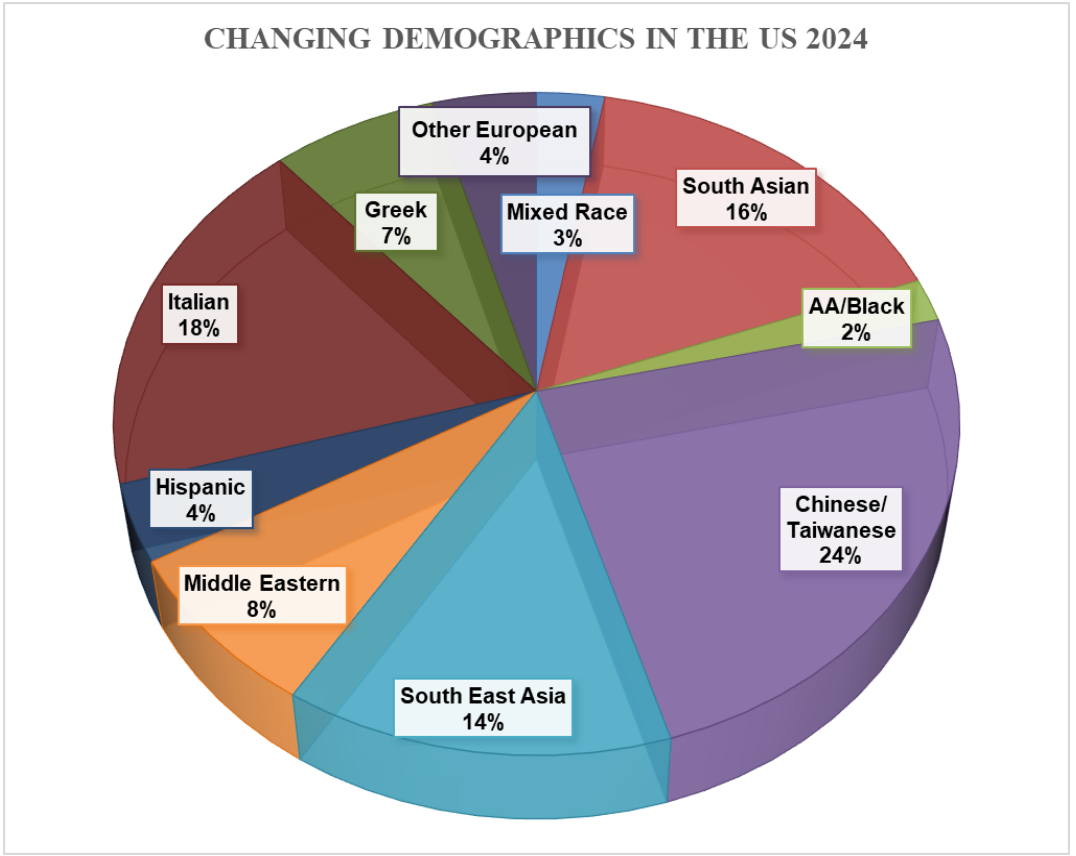
Source: CAF Database

Changes in Database Over 24 years

2001

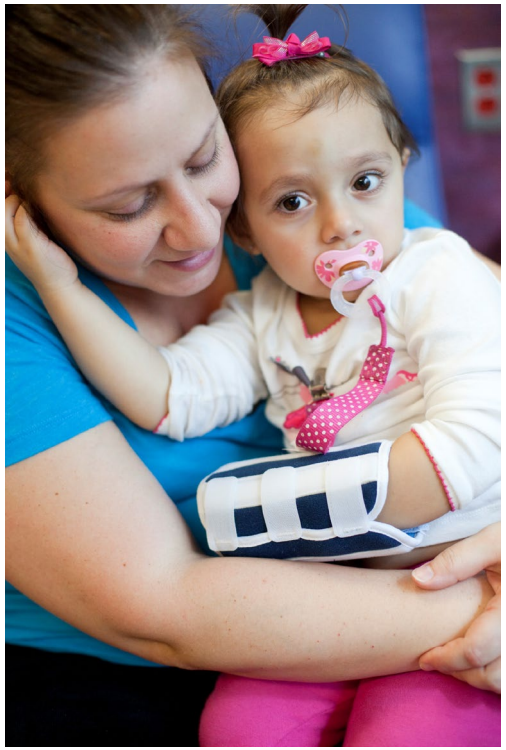
N=690

Majority Italian/Greek descent



Patient/Family Challenges

- Lifelong Treatment – Need for Expert Care
- Time-Intensive Management
- Inconvenience
- Family Dynamics
- Cost/Expense
- Impacts Decisions About Future
  - College
  - Where to Live
  - Career
  - Insurance
  - Marriage/Family
- Cultural/Social Standing



NTDT

Beta Thalassemia Intermedia
E Beta Thalassemia
Hemoglobin h
Hemoglobin H Constant Spring
Other Variants

Great Variation in Severity
Difficult to Identify/Engage Patients
Treatment/Monitoring Needs Vary Significantly

Areas of Opportunity

Barriers to Health Care Access

Innovative Research

Patient Education

Barriers to Health Care Access

Provider Knowledge

Geography

Diagnosis/Reporting

Blood Bank Education

Patient Knowledge

Availability of Chelators

Innovative Research

Freshness of Blood?

Greater Understanding of NTDT Needs

Aging in Thalassemia

Nutrition & Exercise

Patient Education

What Do We Not Know About What Patients Don't Know?
(And Why Don't They Know It?)

Relevance of Information to NTDT Community

Cultural Understanding

Expanding Media Options

Fertility Preservation

Using Administrative Data to Identify Individuals with Thalassemia: Challenges and Opportunities

presented to

The Committee on the Blueprint for Addressing Thalassemia in the United States

April 7, 2025

Surveillance for Rare Non-Infectious Conditions

Public health surveillance according to the World Health Organization (WHO), "the continuous, systematic collection, analysis and interpretation of health-related data needed for the planning, implementation, and evaluation of public health practice.“

Active versus Passive Surveillance

Registries versus Large Administrative Data Sets

Registry and Surveillance for Hemoglobinopathies (RUSH)

- Seven states (California, Florida, Georgia, Michigan, North Carolina, New York, and Pennsylvania).
- In 2008, these seven states represented ~38% of the total population, 42% of the black population, 54% of the Asian population, and 49% of the Hispanic population in the United States (http://www.census.gov).
- Developed administrative case definitions using ICD-9 codes for both Sickle Cell Disease and Thalassemia.
- Accuracy of coding unknown, subtype coding likely inaccurate.
- Linked state-level data from **multiple data sources** from **2004-2008** under public health authority (allows for HIPAA waivers to perform public health surveillance).

State-level Data Sources

- Newborn Screening
  - Screening for SCD has been included on the recommended uniform NBS panel in all 50 states since 2006, but α- and β-thalassemia screening and reporting of results for newborns is currently performed in only a few states.
- Clinical Case Reporting (by Site)
- Medicaid Administrative Claims Data
- Hospital Discharge Data
- Vital Records Data (not used for case finding)
- Department of Public Health likely not the data steward for all data sources required data sharing agreements

RUSH: Thalassemia Case Definition

Level 1 (Confirmed): CLIA-certified laboratory result of DNA mutation analysis for thalassemia reported by a state newborn screening program with confirmatory testing **OR** Clinical diagnosis by a physician with documented confirmatory CLIA-certified laboratory testing by DNA mutation analysis after the newborn period.

Level 2 (Probable): CLIA-certified laboratory result of thalassemia reported by a state newborn screening program without DNA mutation analysis **OR** Clinical diagnosis by a physician with documented confirmatory CLIA-certified laboratory testing but without DNA mutation analysis after the newborn period **OR** Thalassemia ICD code at two or more separate health-care encounters PLUS one or more thalassemia-associated complication, treatment, or procedure.

Level 3 (Possible): CLIA-certified laboratory result of thalassemia reported by results of state newborn screening program without report of confirmatory testing **OR** Thalassemia ICD code for a single health-care encounter.

Case Definition Codes

Diagnosis Codes (no separate code for trait prior to 2012):

- 282.4 Thalassemias
- 282.49 Other thalassemia
- 282.7 Other hemoglobinopathies

Complications: Pulmonary hypertension, iron overload, gall stones, cholelithiasis, cholecystitis, splenomegaly, splenic sequestration or hypersplenism

Treatments: Iron chelators

Procedures: Red cell transfusion, splenectomy, cholecystectomy

Results

Table 3 Number of individuals with a thalassemia diagnosis by state and case definition level, 2004–2008

State	Level 1	Level 2	Level 3
California	836	1,915	14,534
Florida	14	1,620	6,731
Georgia	64	271	4,553
Michigan	6	31	1,354
New York	33	1,873	10,380
North Carolina	84	232	1,127

*307 confirmed cases linked to Medi-Cal claims data

Public Health, Research, Epidemiology and Surveillance in Hemoglobinopathies (PHRESH)

- Both states found that the RuSH methodology for identifying and determining an approximate case count as well as investigating health care utilization for SCD was appropriate using Level 1 & Level 2 definitions. Further studies reported sensitivity, specificity, and PPVs for the confirmed & probable definition and updated and simplified administrative definitions.
- Challenges occurred with the thalassemia definition, initial methodology overcounted cases (no separate ICD-9 code for trait); revised methods tested by CA appeared to undercount cases.

California PHRESH Results

	Level 1: Confirmed	Level 2: Probable	Level 3: Possible	Total
Thalassemia	836	1,915	14,534	17,285
Sickle Cell Disease	1,976	3,149	8,724	13,849
Mixed	N/A	559	385	944

- Smaller disease population, more susceptible to identification errors
- Almost 1/3 of probable thalassemia cases had mixed diagnosis codes
- Of the 1,915 cases meeting the probable case definition, nearly all 1,789 were adults and nearly half of those adults (840) were 70 years of age or older. This age distribution was not consistent with expectations for the disease population only 2% over age 69 and one would expect higher numbers in the 20-40 age group.
- 12% of the adult cases are Hispanic and 28% are Asian which also doesn't match the confirmed adult thalassemia cases 3% Hispanic and 64% Asian.
- Suspected a number of false positives.

PHRESH Findings-Thalassemia challenges

- Less frequently hospitalized or seen in the ED (42% of Level 1 SCD, 17% of Level 1 Thalassemia)
- Less likely to be covered by the State Medicaid program (36% of Level 1 Thalassemia vs. 56% of Level 1 SCD); miss those privately insured
- Smaller diseased population, more susceptible to identification errors (836 confirmed thalassemia cases vs. 1,976 confirmed SCD cases)
- Few states screen for thalassemias via NBS and many people were not born in the US or were born prior to the initiation of universal NBS
- Lower sensitivity of definitions when there are few healthcare encounters (equal numbers of true positives and false negatives even in SCD population)

Thalassemia Opportunities

- Suggest separate definitions for Thalassemia and SCD surveillance
- California Genetic Disease Screening Program (GDSP) began screening for beta thalassemias in 1990 and clinically significant alpha thalassemias in 1999
- 307 confirmed cases found in MediCal administrative data
- Accuracy (PPV) of case definitions could be improved with the addition of transfusion and iron overload codes
- ICD-9 codes updated in 2012 and a trait code for thalassemia was added

Updated ICD-9/10 codes

ICD-9 codes

282.4	Thalassemias
282.49	Other thalassemia
282.7	Other hemoglobinopathies
*282.40	Thalassemia, unspecified
*282.43	Alpha thalassemia
*282.44	Beta thalassemia
*282.45	Delta-beta thalassemia
*282.47	Hemoglobin E-beta thalassemia

***ICD9 codes expanded for THAL in 2012 ICD9 version**.*

ICD-10 codes implemented in 2016

D56.0	Alpha thalassemia
D56.1	Beta thalassemia
D56.2	Delta-beta thalassemia
D56.4	Hereditary persistence of fetal hemoglobin [HPFH]
D56.5	Hemoglobin E-beta thalassemia
D56.8	Other thalassemias
D56.9	Thalassemia, unspecified

Angela Snyder

Research Professor

angiesnyder@gsu.edu

Georgia Health Policy Center

55 Park Place NE, 8th Floor
Atlanta, GA 30303
ghpc.gsu.edu

CDPH
California Department of Public Health

Newborn Screening for Hemoglobinopathies in California

Rajesh Sharma, PhD, FACMG
Laboratory Director
Genetic Disease Screening Program (GDSP)
California Department of Public Health

Objectives

- Describe California's approach to screening and confirmatory testing
- Numbers of confirmed cases identified
- Future directions

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GDSP Mission

- Ensures all pregnant women in California can access voluntary prenatal screening for specific fetal birth defects.
- Screens all babies born in California for serious but treatable genetic disorders to prevent or minimize adverse outcomes through early detection and management of disease.
- The California Newborn Screening (NBS) Program screens approximately 410,000 babies annually for 35 primary disorders and 54 secondary disorders, and about 300 babies are found to have severe disorders.

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Criteria for Newborn Screening

- Important health problem
- Markers associated with disease
- Amenable to treatment
- Reliable, economical screening test available
- Adequate methods of confirmation and follow-up available

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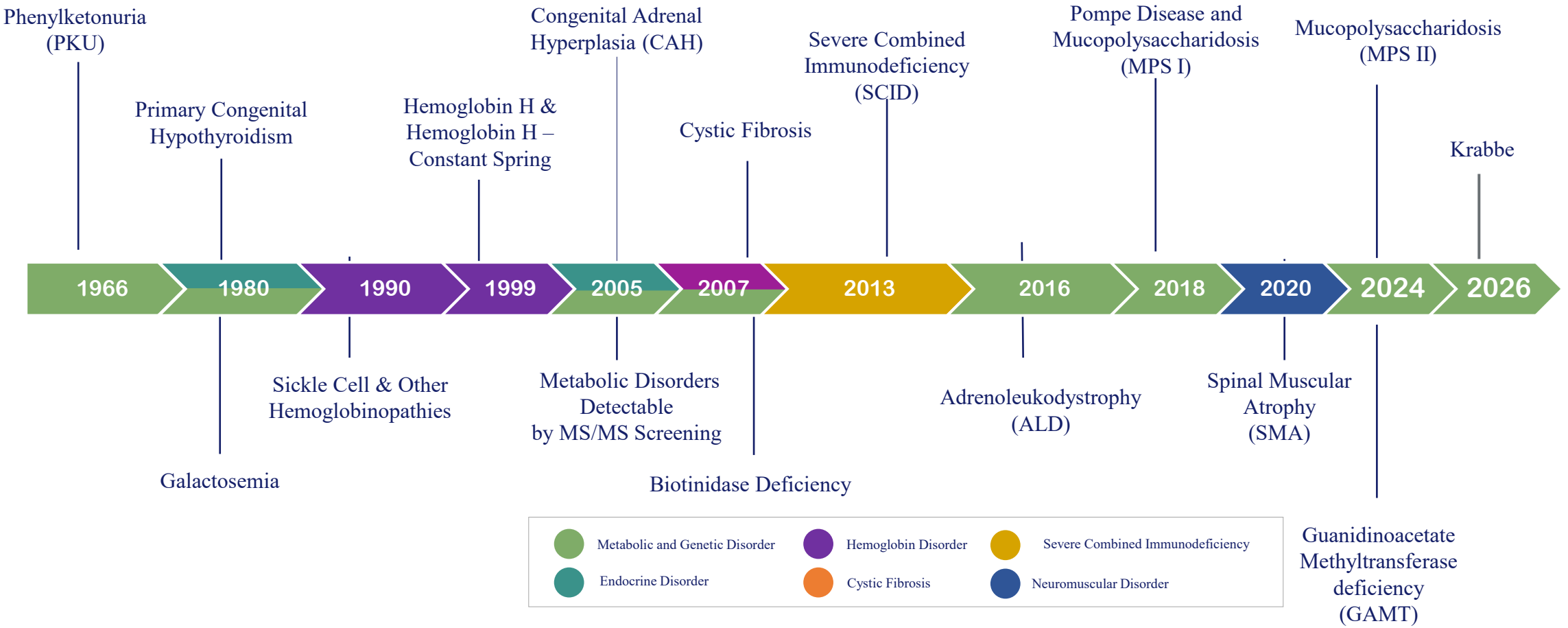
Conditions Screened

Advancements in the California Newborn Screening Program (1966-2024)



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Screening for Hemoglobin Disorders in California

- Alpha Thalassemias: Hb Bart's syndrome (hydrops fetalis), Hemoglobin H disease
- Beta Thalassemias: Major, Hb C Beta Thal, Hb S Beta Thal Hb D Beta Thal, Hb E Beta Thal, Hb E Delta-Beta Thalassemia, HB variant/Beta Thal,
- Beta hemoglobin variants: Hb C, Hb D, HB E, Hb SD, Hb SE, Hb S + variant, and Hb variant plus variant
- Hereditary persistence of Fetal Hb: HPFH/HPFH, S/HPFH

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Hemoglobin Types

- Hemoglobin is tetrameric protein in red blood cells, composed of four globin subunits – two alpha-like globin chains and two beta-like globin chains working in conjunction with heme to transport oxygen in the blood.
- Hb F (α2γ2), the predominant hemoglobin throughout fetal life, constitutes approximately 70% of total hemoglobin at birth.
- The predominant adult human hemoglobin, Hb A (α2β2), constitutes approximately 95-98% of total hemoglobin.

Stage in Development	Hemoglobin	Structure
Embryonic	Gower-I Gower-II Portland-I	$\zeta_2\varepsilon_2$. $\alpha_2\varepsilon_2$. $\zeta_2\beta_2$
Fetal	F	$\alpha_2\gamma_2$
Adult	A A_2	$\alpha_2\beta_2$ $\alpha_2\delta_2$

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Hemoglobin Variants

- More than a thousand genetic variants are known to affect globin chains.
- Variants that result in qualitative changes, such as amino acid substitutions that alter hemoglobin function, cause hemoglobinopathies including HbS, HbC, HbD, HbE and HbG.
- Variants that result in decreased production of globin chains cause thalassemias.

Alpha and Beta Thalassemia

- In alpha thalassemias, absent or diminished synthesis of alpha-globin chains leads to an excess of beta chains (or, in the fetus, of gamma chains) that can form tetramers and trigger hemolysis in more severe forms.
  - Hb Bart syndrome (hydrops fetalis) (most severe)
    - No alpha-globin produced
    - Usually results in stillbirth or death during the neonatal period
  - Hemoglobin H disease
    - Reduced alpha-globin levels
    - Has a broader phenotypic spectrum but can cause serious disease
- In beta thalassemias, excess alpha globin chains accumulate, forming aggregates that can injure the blood cell membrane and lead to hemolytic anemia.

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Hemoglobinopathy Screening & Diagnosis

- In California, routine newborn screening for hemoglobinopathies is accomplished by high-performance liquid chromatography (HPLC).
- 200 newborns/year are identified with a possible Hb condition.
- In addition, we identify newborns with Hb trait.
- Molecular confirmatory testing is performed at UCSF Benioff Children's Hospital in Oakland, arranged and paid for by our program.

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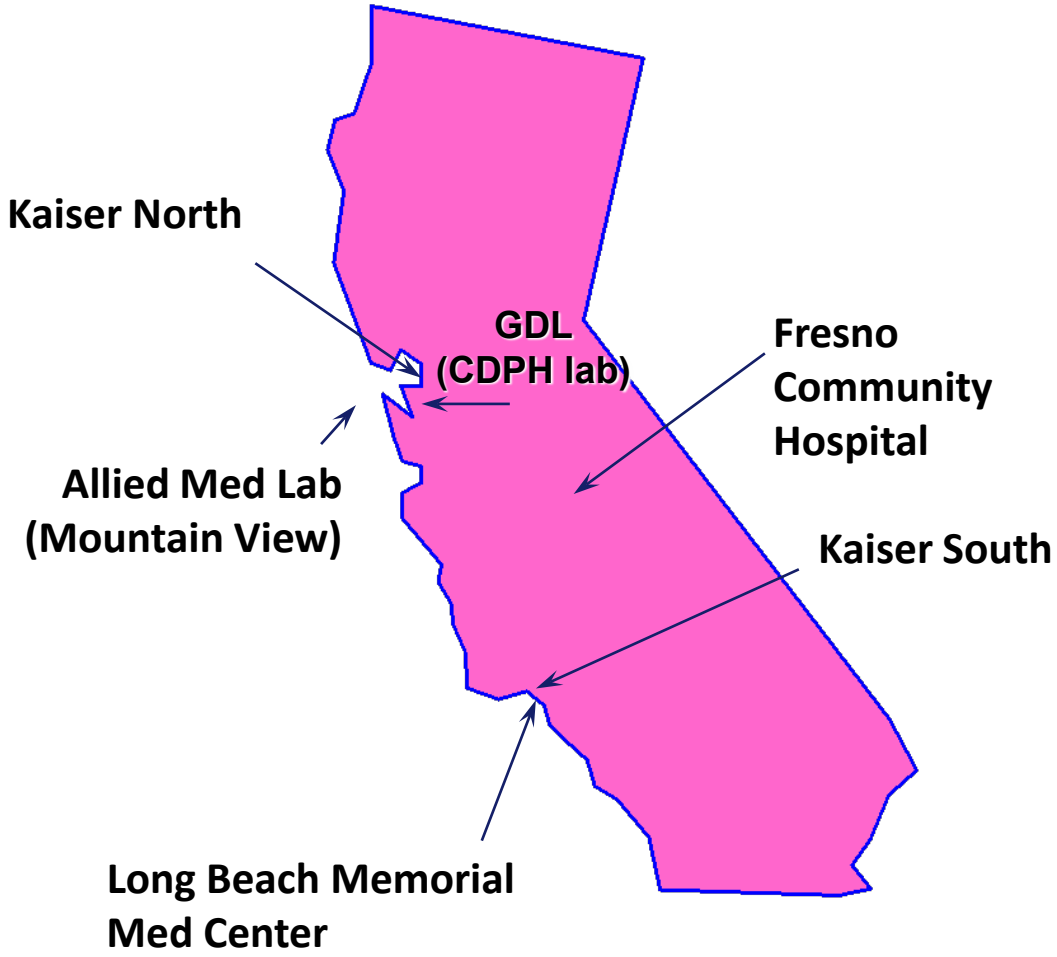
California NBS Laboratory Activities

- Initial specimen processing, data entry, and most first-tier screening are done at contracted regional labs.
- GDL is involved in all biochemical screening processes including QA/QC, with regional results reviewed, interpreted, and released by GDL staff.



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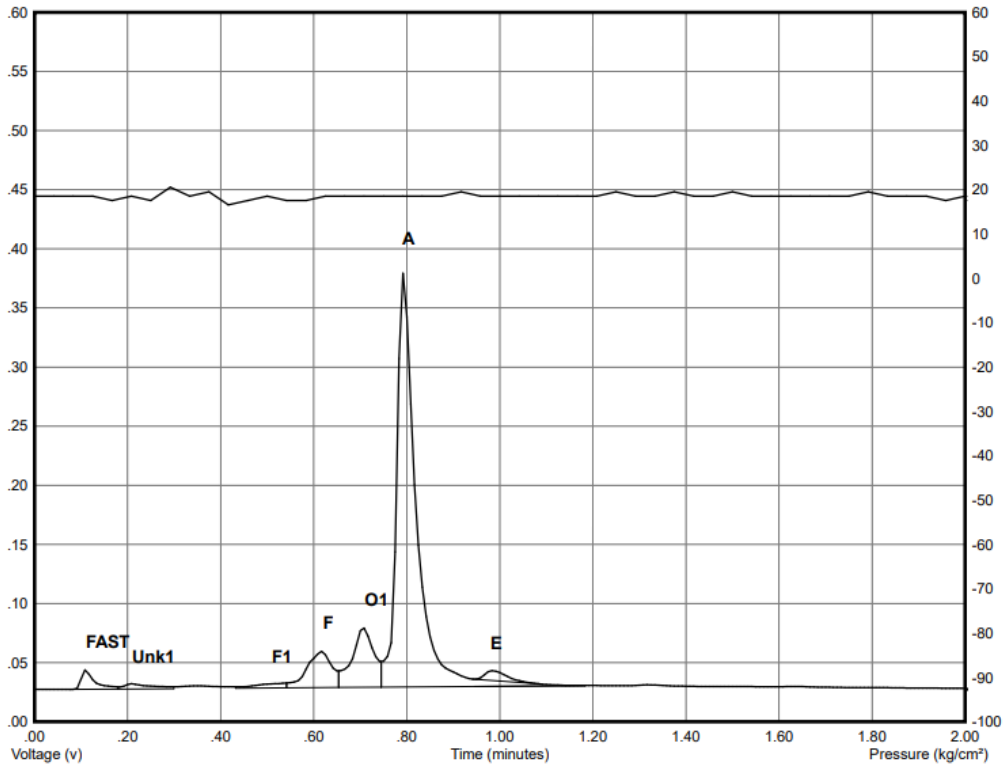
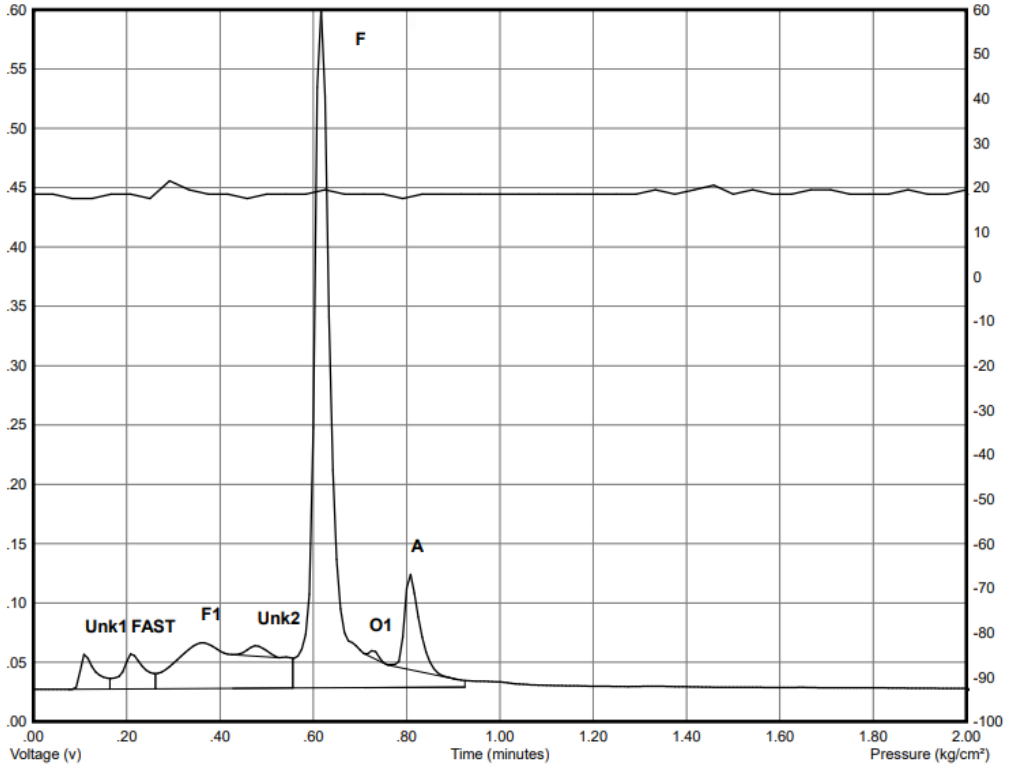
HPLC Normal Hemoglobin Pattern

Normal Adult



Normal Newborn



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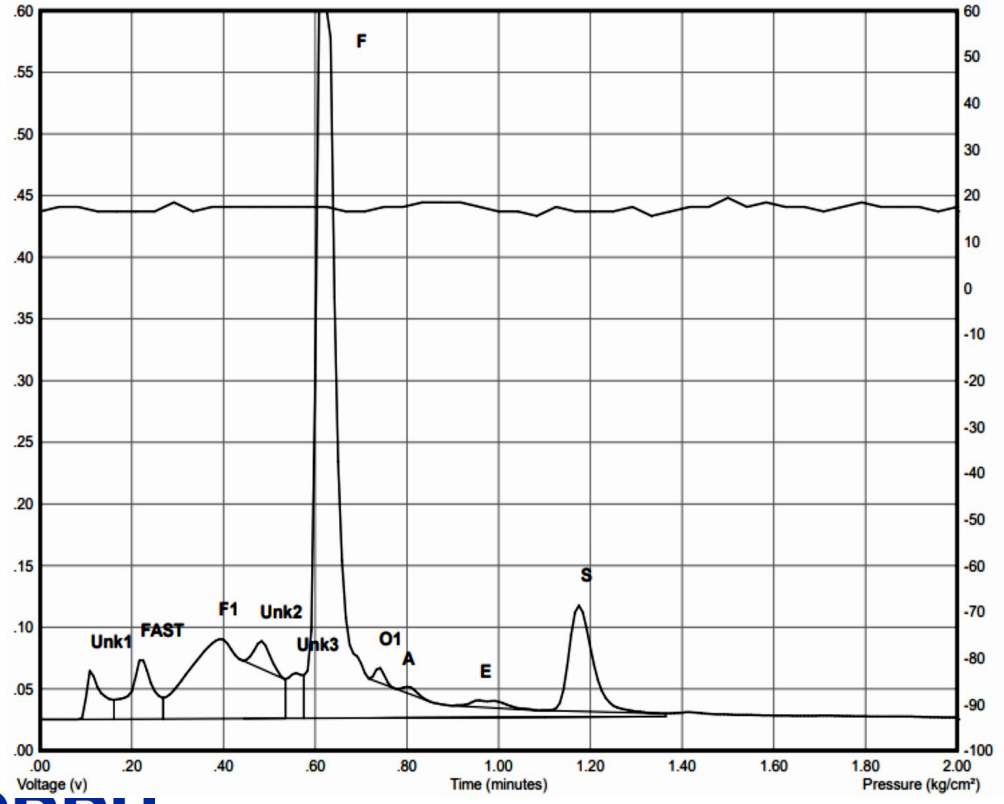
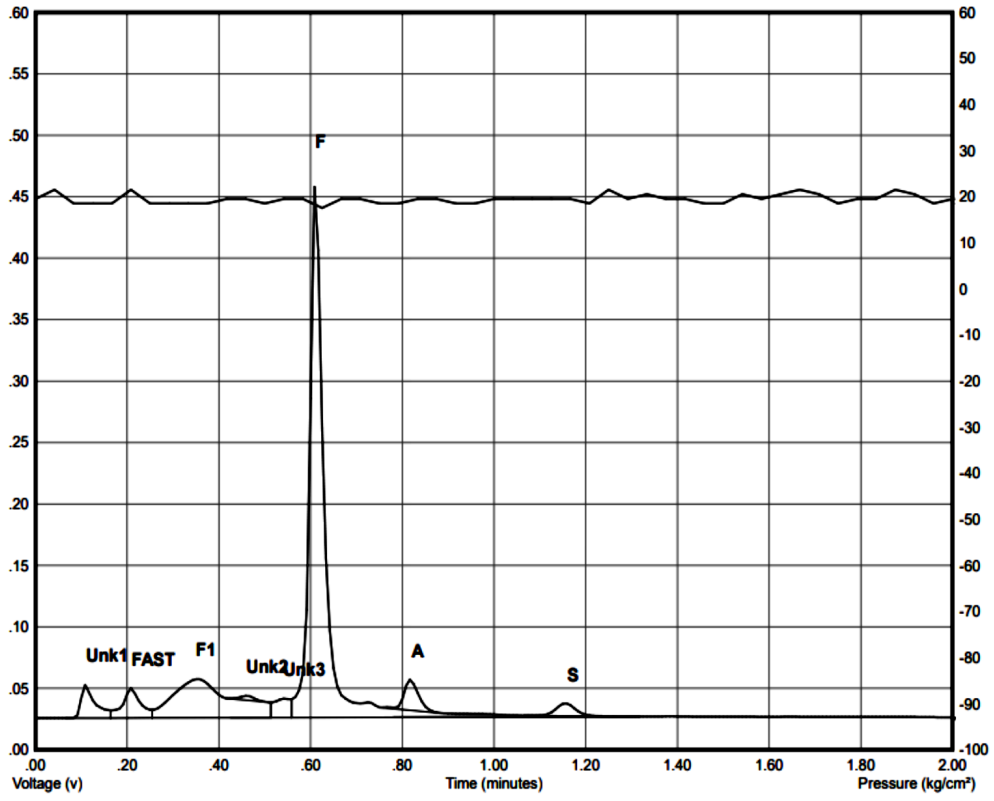
HPLC Abnormal Hemoglobin Pattern

Sickle Cell Disease



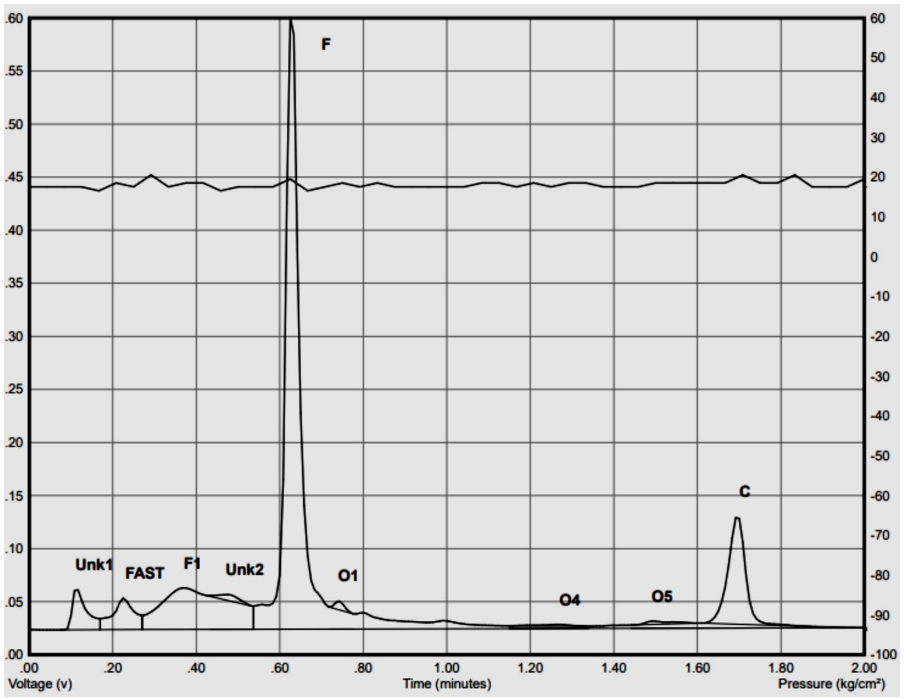
Sickle Cell Trait



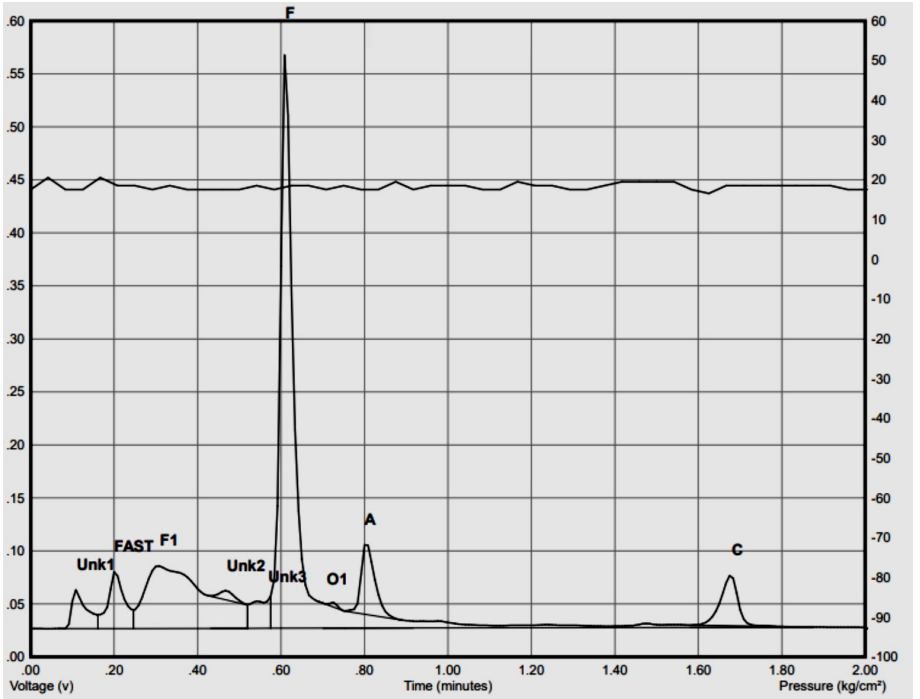
CDPH

HPLC Abnormal Hemoglobin Pattern

Hemoglobin C Disease



Hemoglobin C Trait



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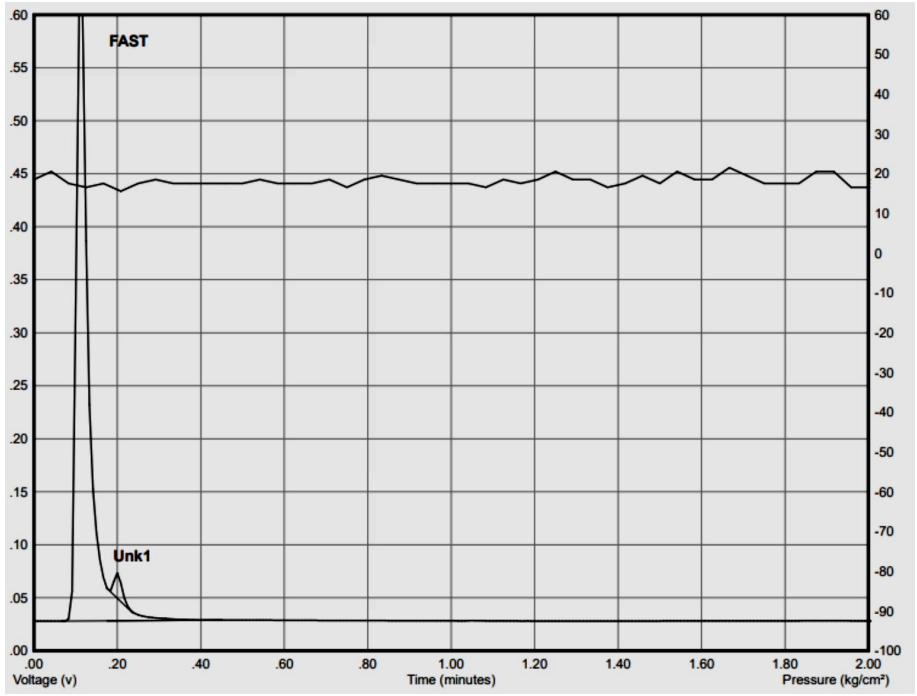
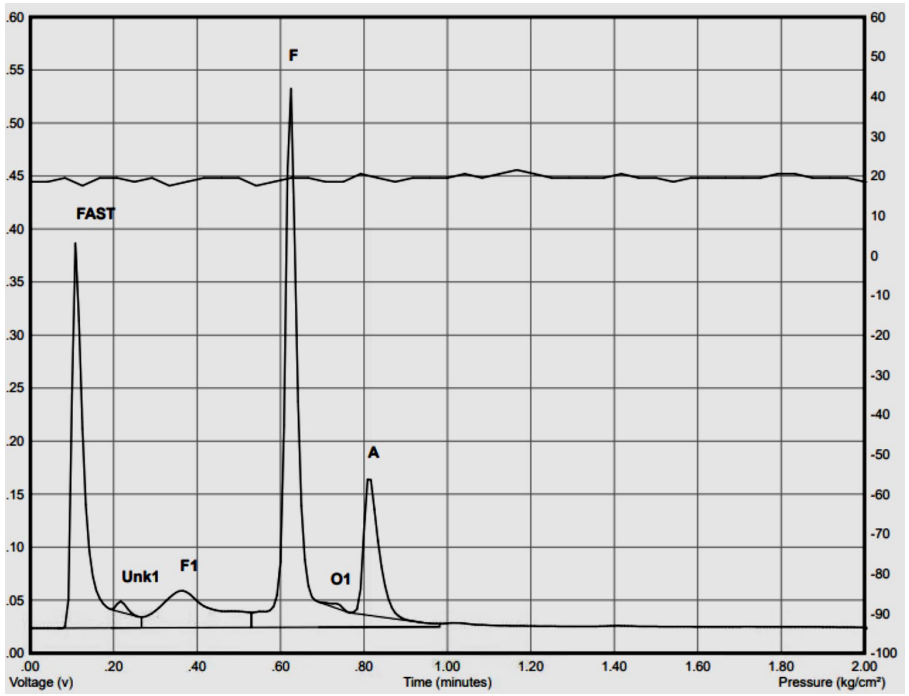
HPLC Abnormal Hemoglobin Pattern

Alpha Thalassemia Major



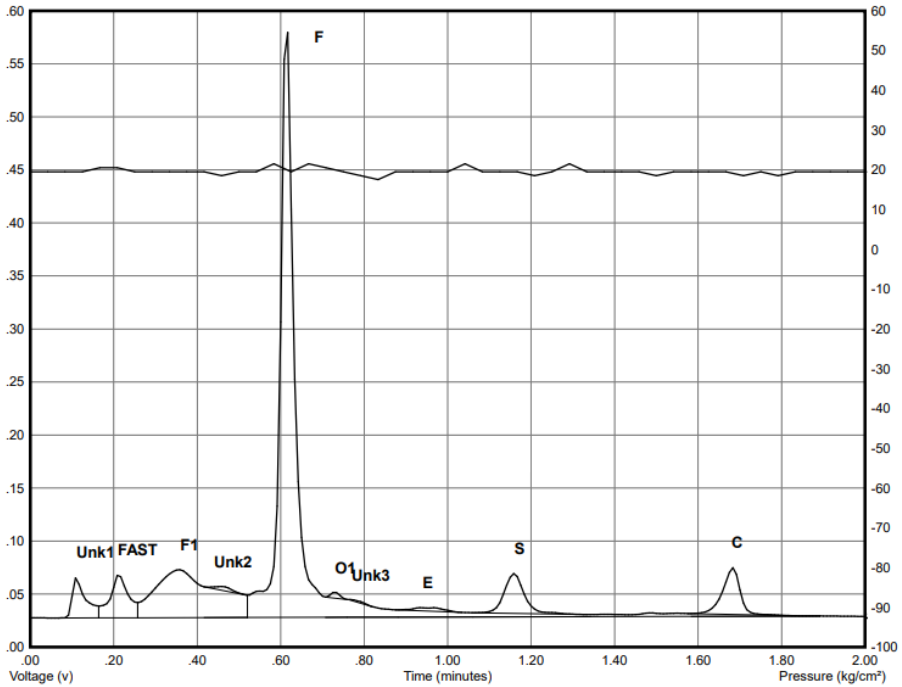
Hemoglobin H Disease



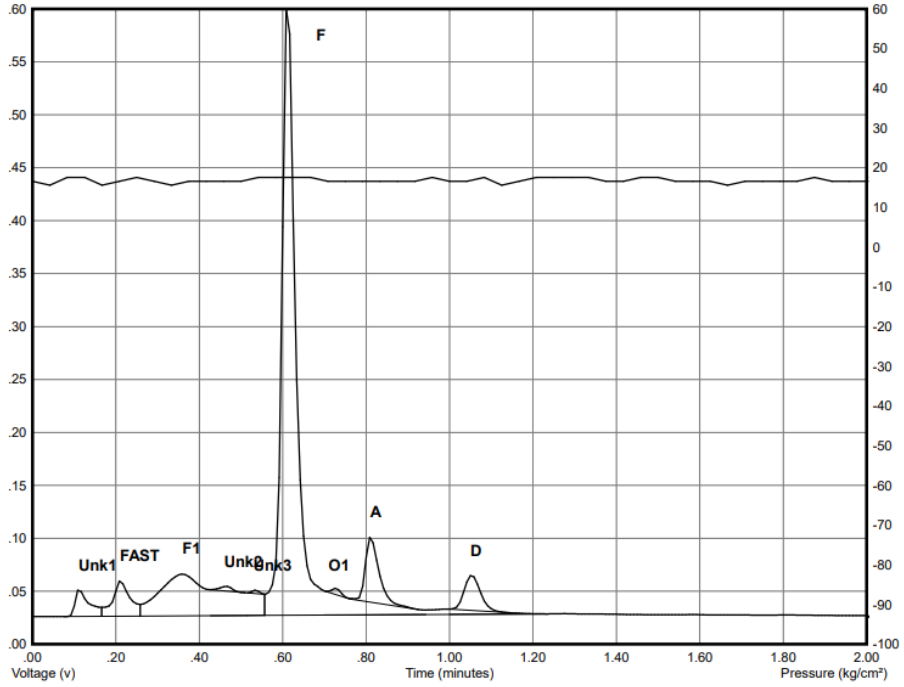
CDPH

HPLC Abnormal Hemoglobin Pattern

Hemoglobin SC Disease



Hemoglobin D Trait



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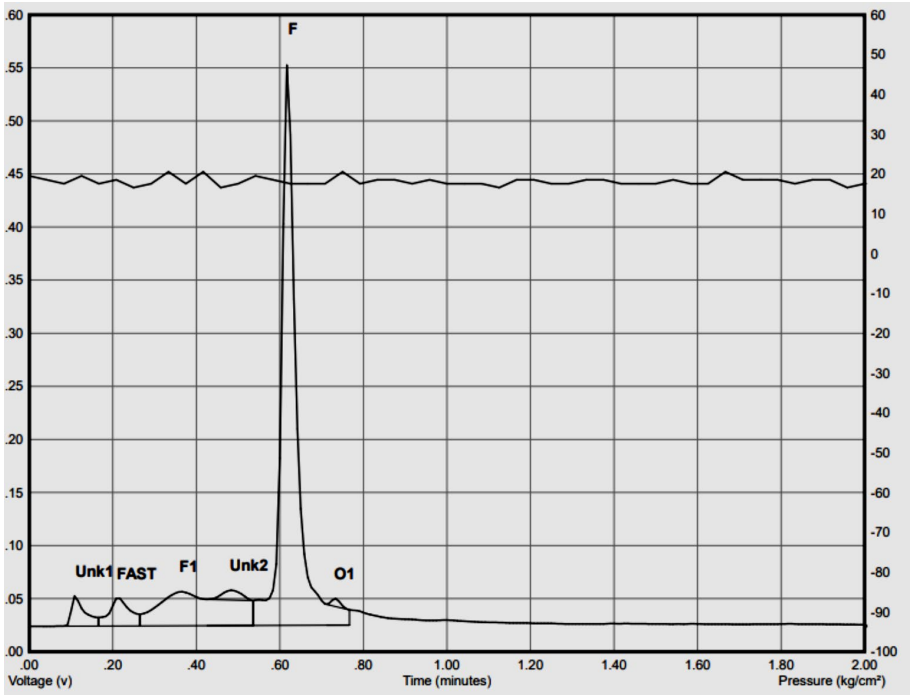
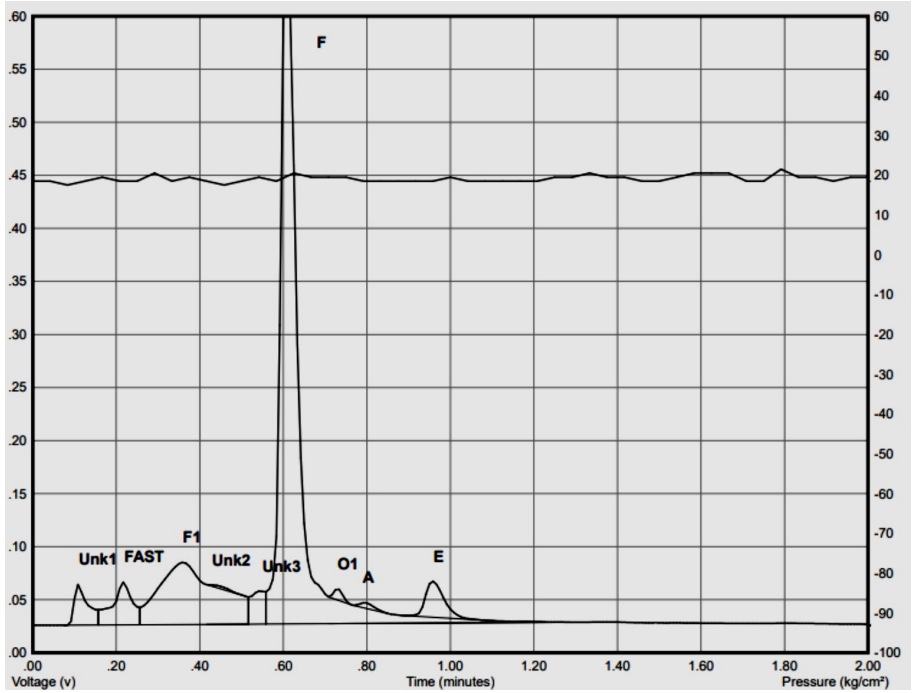
HPLC Abnormal Hemoglobin Pattern

Beta Thalassemia Major



Hemoglobin E Disease



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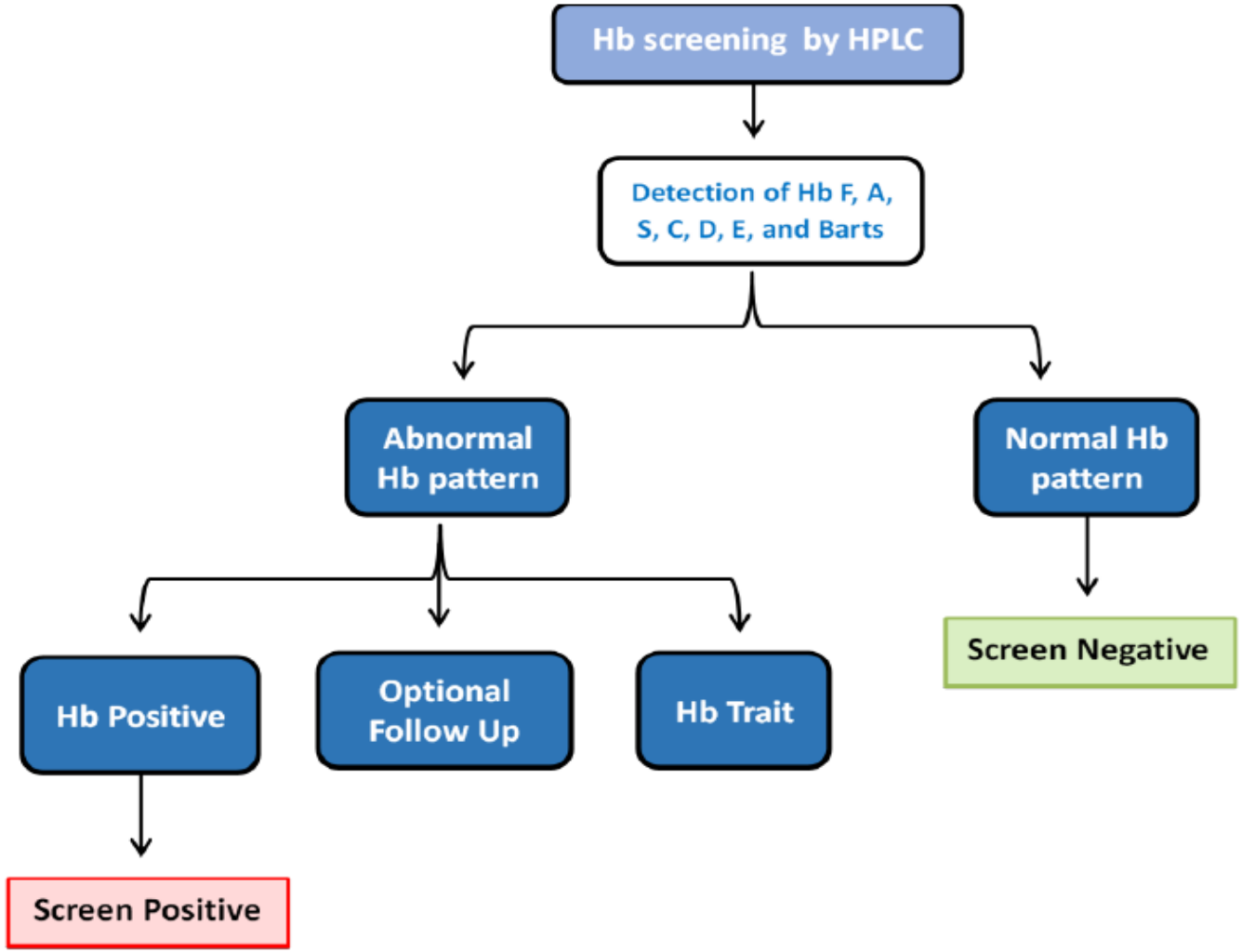
California Follow-up Protocol



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Top 10 Hemoglobinopathy-Positive Cases (January 2005 – March 2025)

Name	Number
Hb S/S (Sickle S/S disease)	899
Hemoglobin H Disease	792
Hemoglobin E Disease	751
Hb S/C (Sickle S/C disease)	497
Hemoglobin C Disease	89
Variants (homozygous or heterozygous)	84
Hemoglobin H/Constant Spring Disease	57
Hb S/E (Sickle S/E disease)	56
Beta Thalassemia Major	52
Alpha Thalassemia Major	24

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California Hemoglobin Trait Follow-up Program

2022	
S Trait	3389
C Trait	807
D Trait	241
Total	4437

- These are the three most common and medically important Hb variants detected.
- Of ~410,000 births/year in California, 1 in 95 will be identified with one of these traits.

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California Hemoglobin Trait Follow-up Program

National consensus around the development of NBS for sickle cell disease stressed the importance of Hb trait status to reproductive decision making in families.

Future Directions

- Staff Training: Creating a library of abnormal peaks for teaching purposes
- Performing in-house molecular sequencing for hemoglobinopathies as second-tier screening.

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