Haemoglobinopathy Prevention

Global strategies, UK approach and future directions

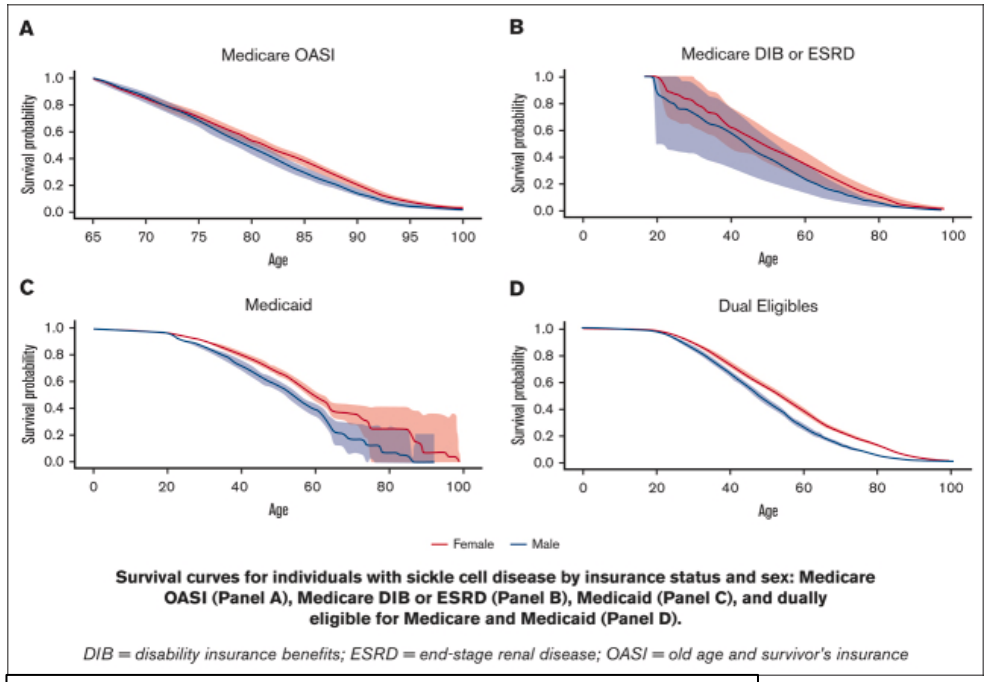
Dr Clare Samuelson

Consultant Haematologist

The Case for Prevention

Definition: Preventing birth of babies affected by clinically significant haemoglobinopathies

Life Expectancy Remains Short in Sickle Cell Disorder



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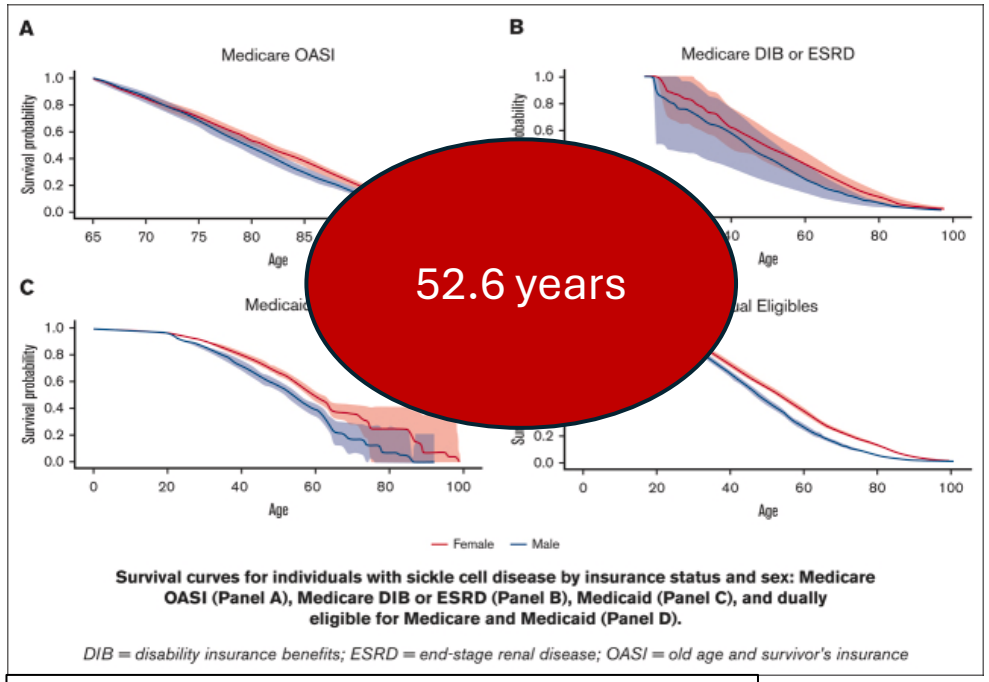

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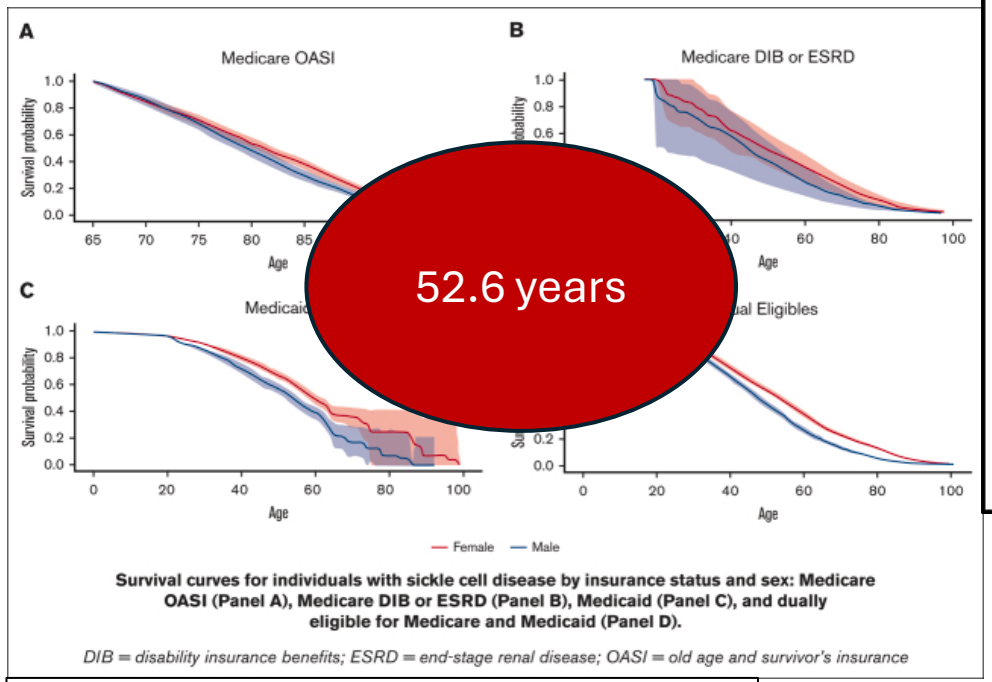

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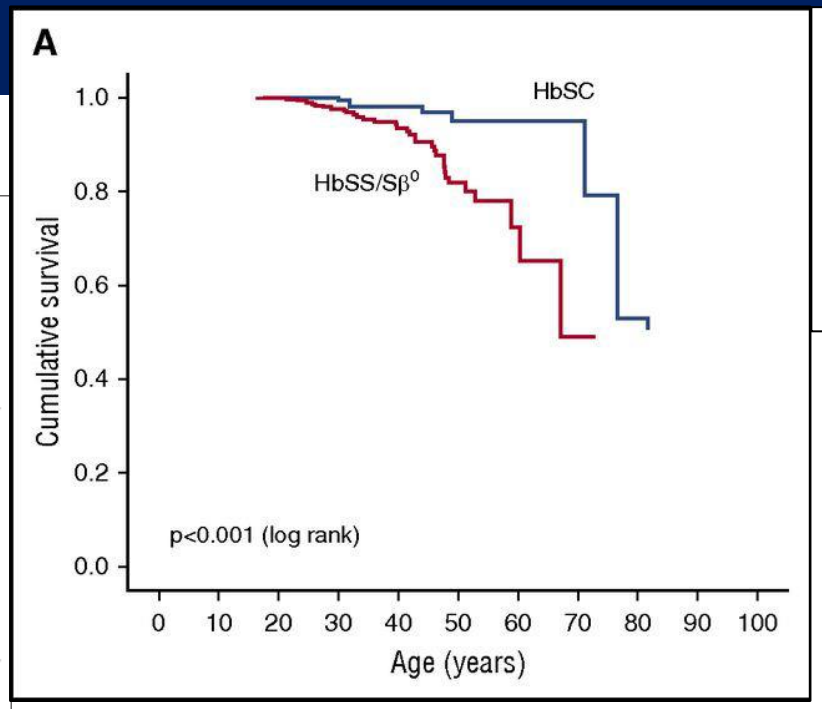

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Volume 128, Issue 10, 8 September 2016, Pages 1436-1438

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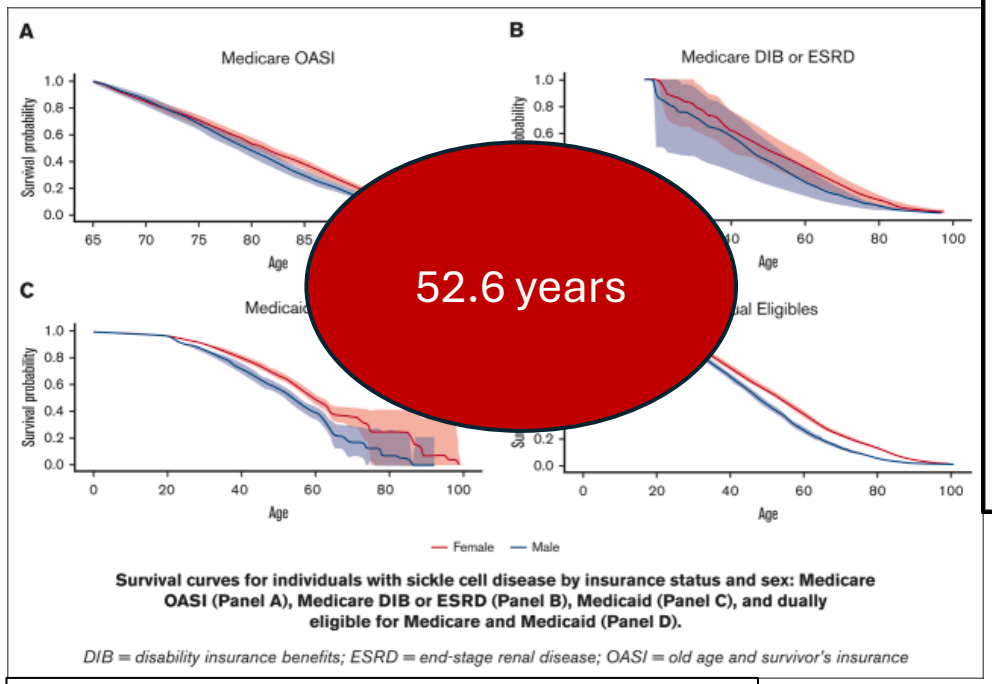

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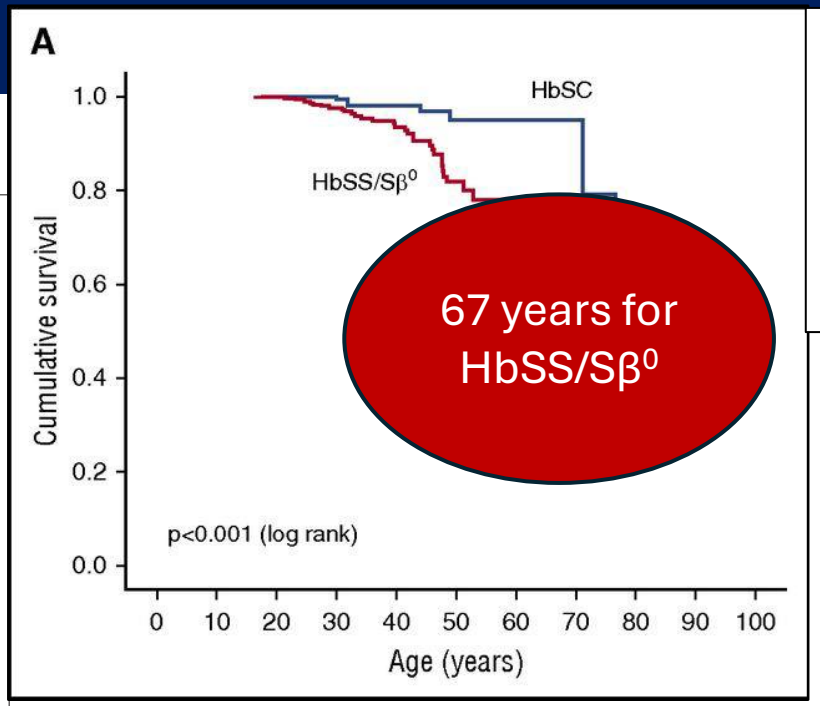

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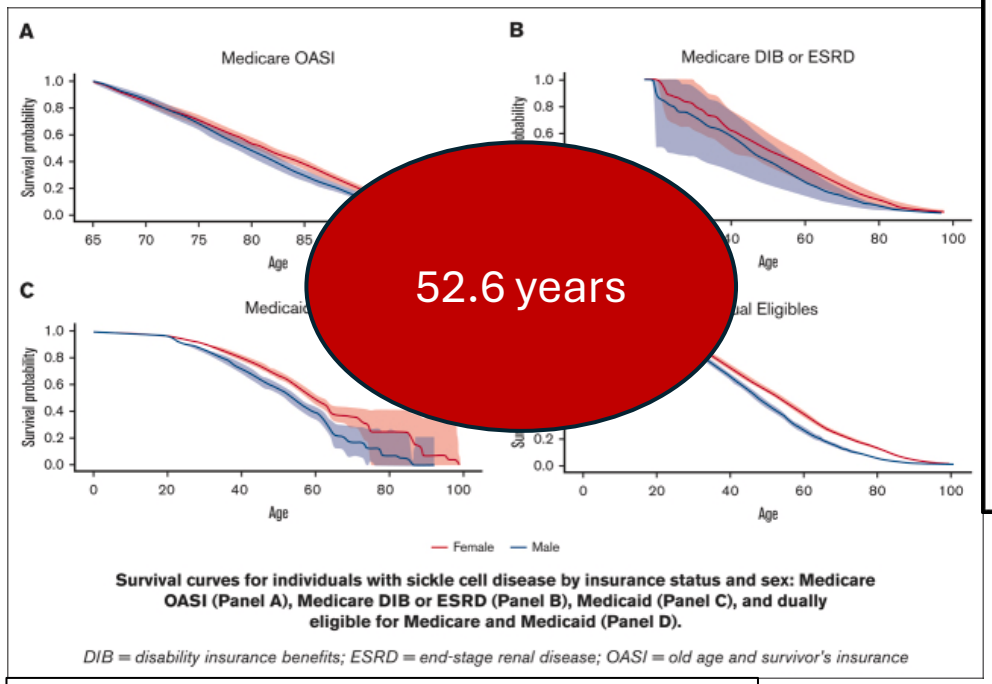

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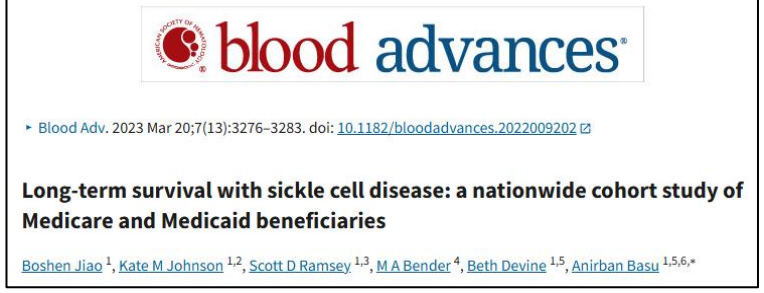
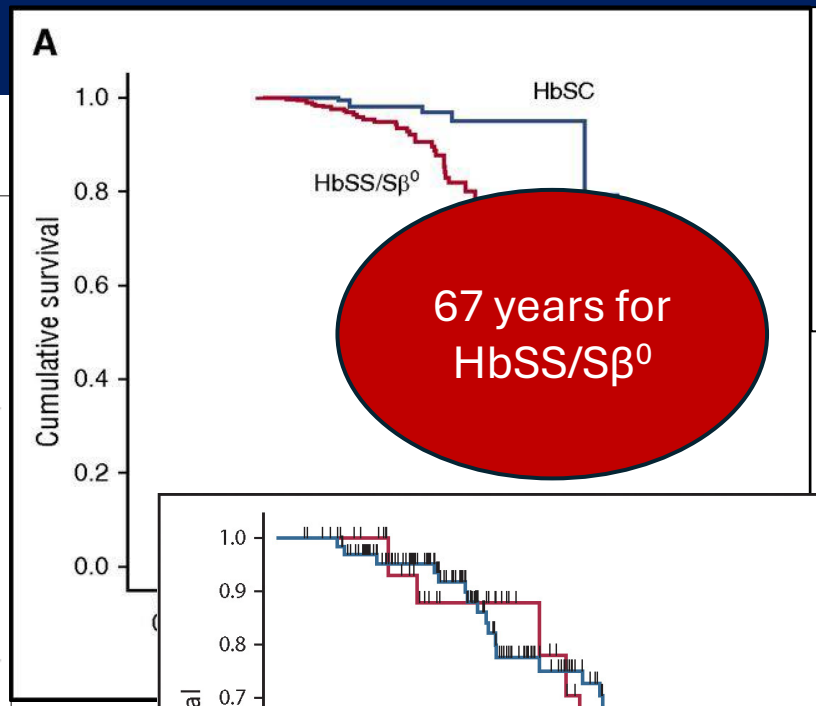
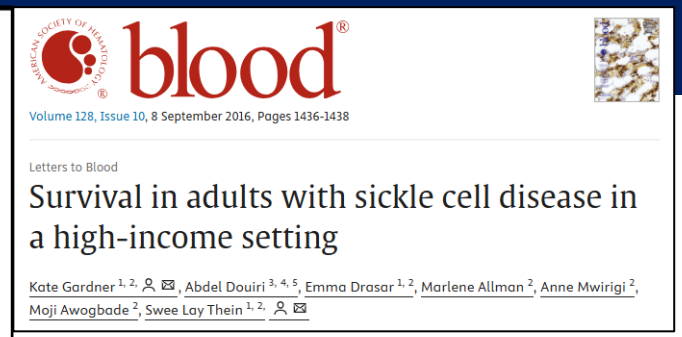
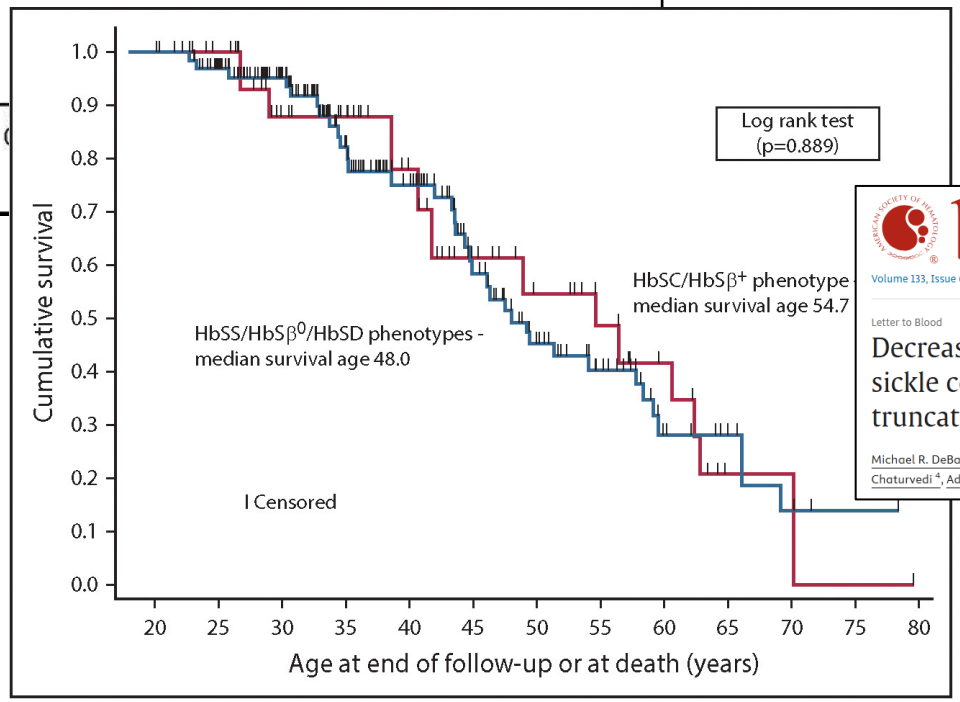
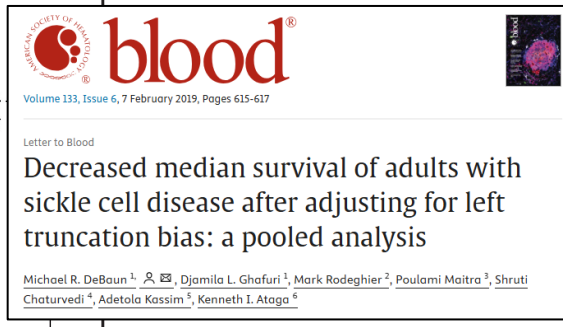




67 years for HbSS/Sβ^0







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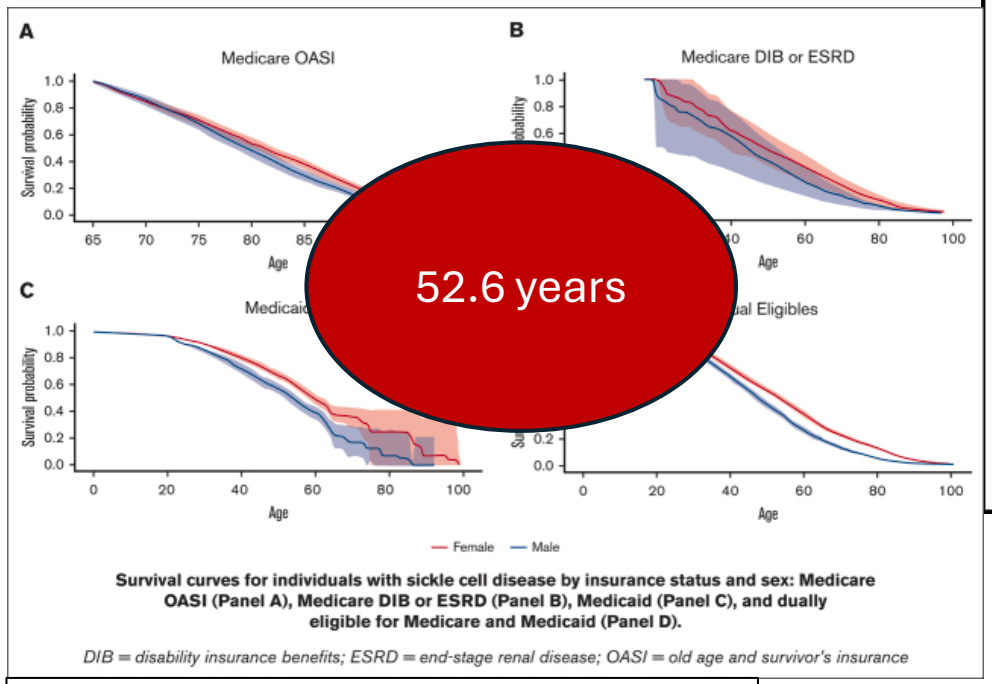

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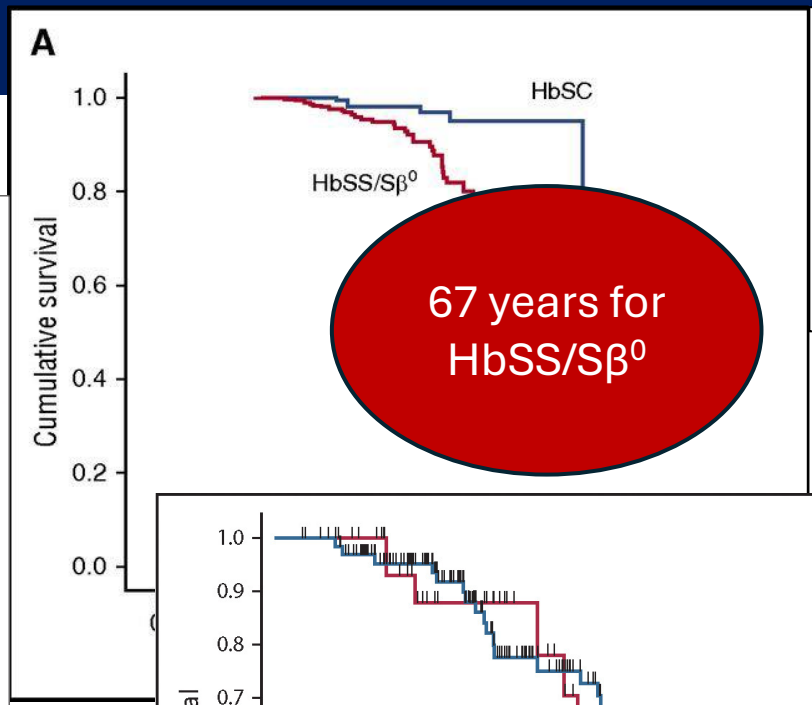

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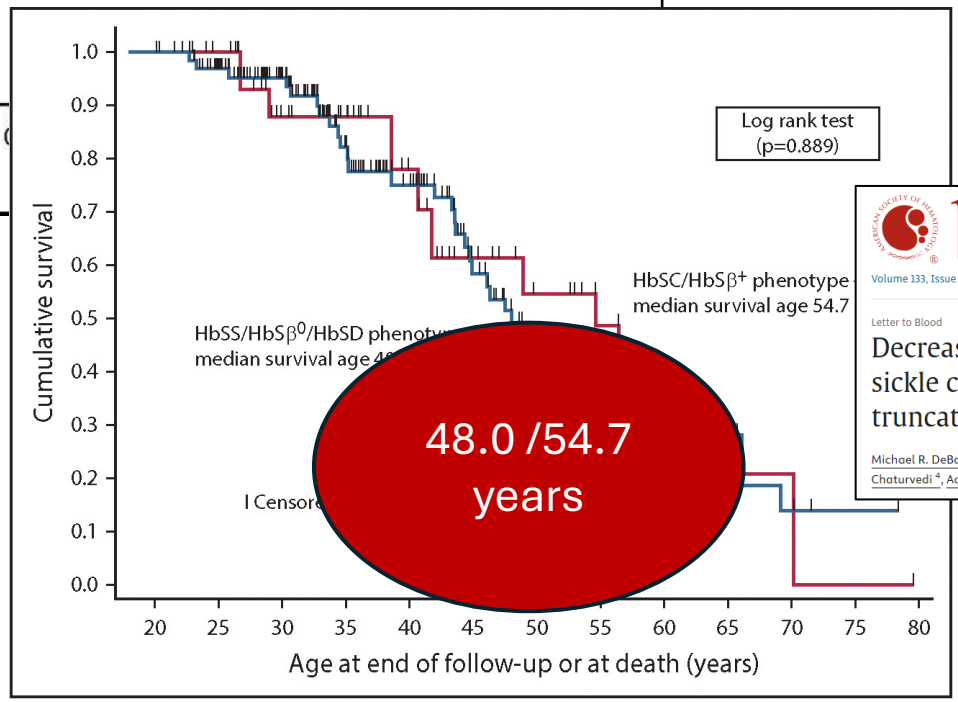

48.0 /54.7 years

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Michael R. DeBaun [1], Djamila L. Ghafuri [1], Mark Rodeghier [2], Poulami Maitra [3], Shruti Chaturvedi [4], Adetola Kassim [5], Kenneth I. Ataga [6]

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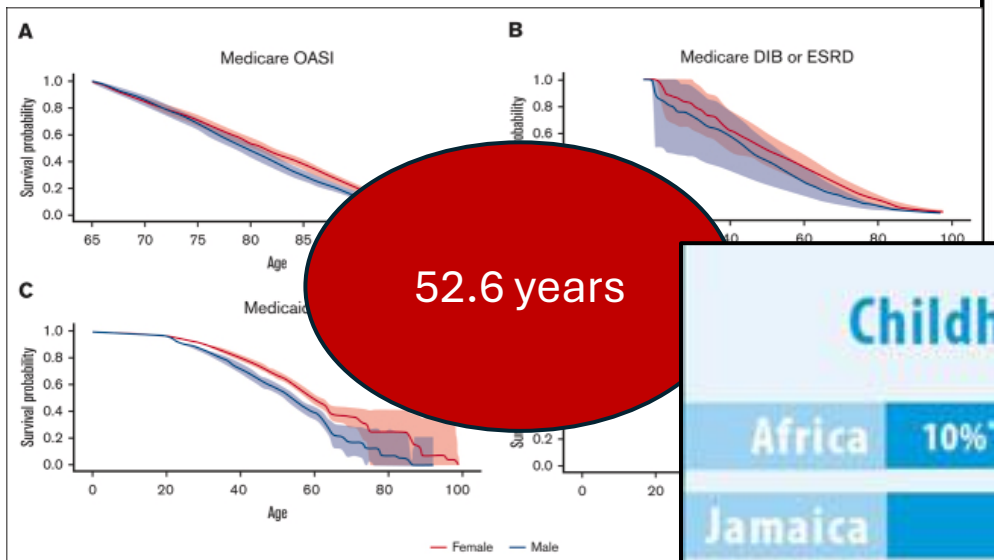

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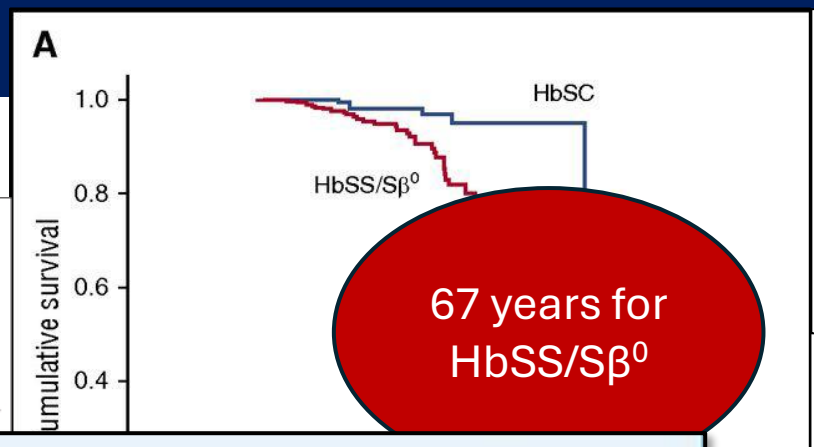

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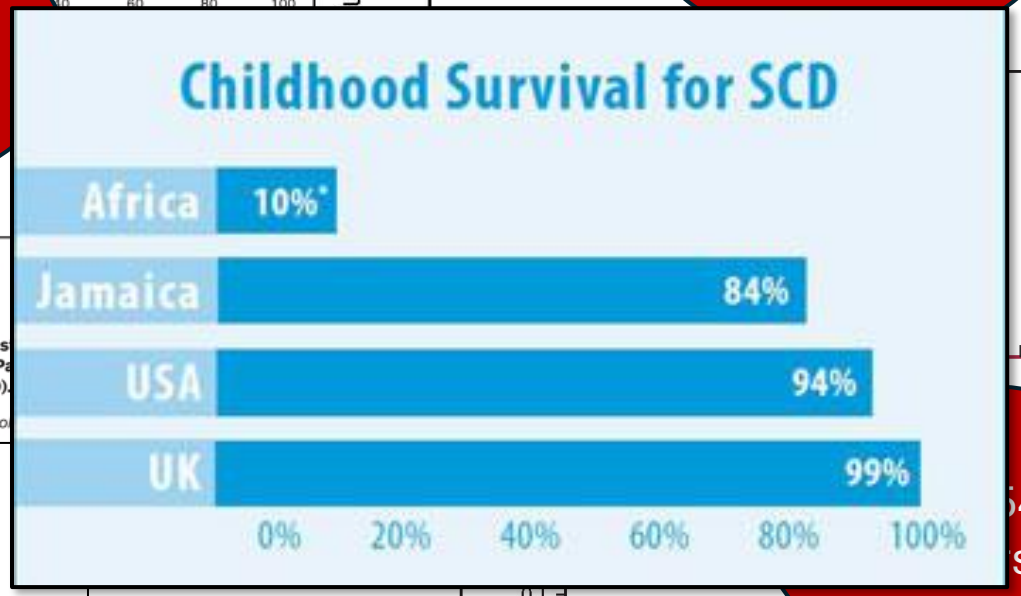
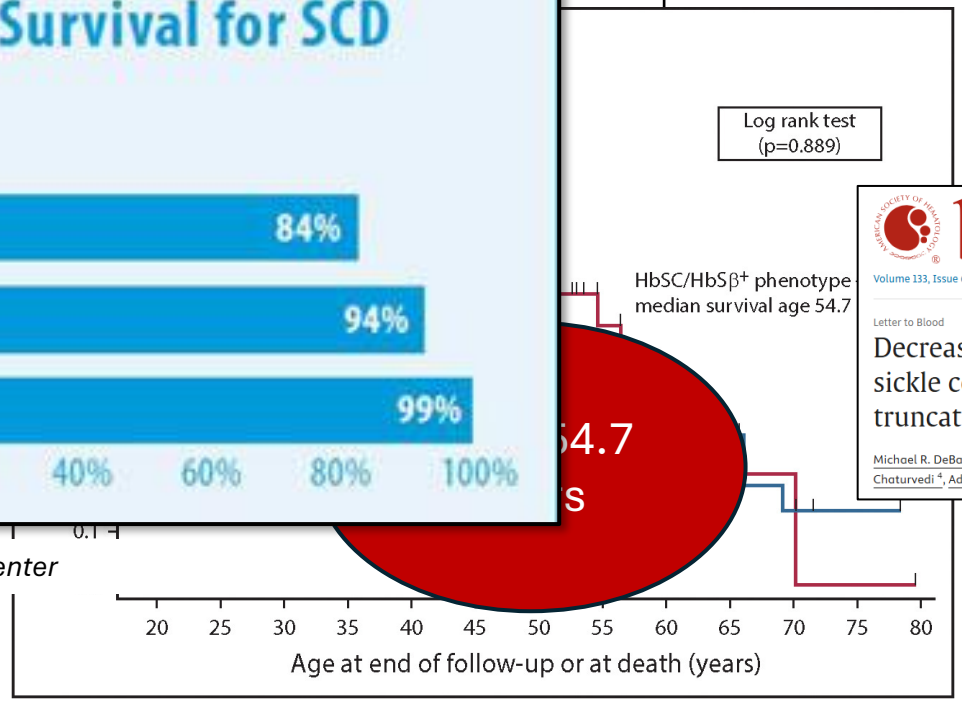


NIH, Fogarty International Center



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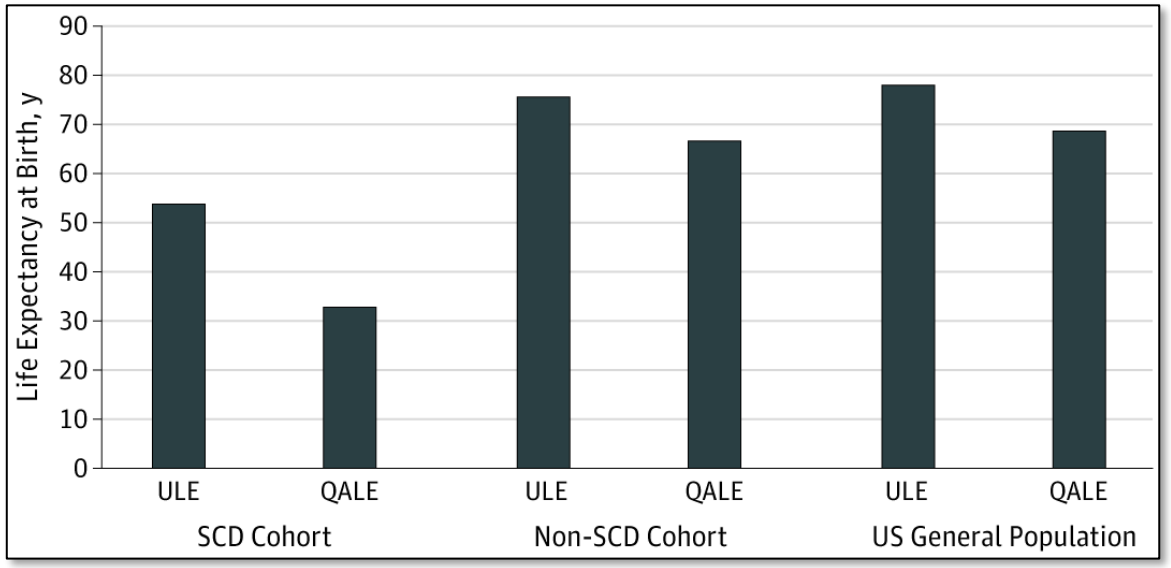
Poorer Quality of Life in Sickle Cell Disorder



Expected Life-Years and Quality-Adjusted Life-Year Decrements by Age

Sickle cell disease (SCD), QALE: quality-adjusted life expectancy; ULE: unadjusted life expectancy.

Lubeck et al, JAMA Netw Open, 2019

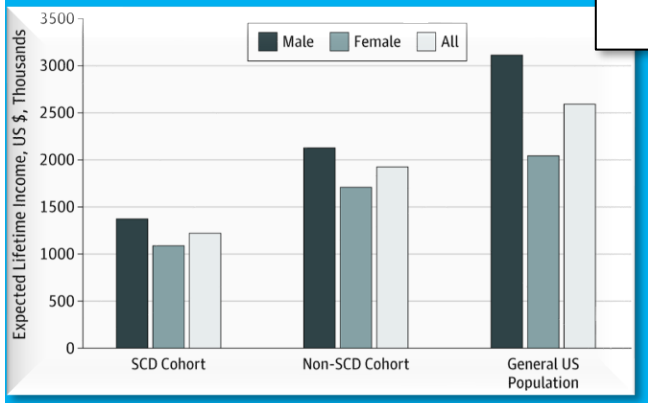
Poorer Quality of Life in Sickle Cell Disorder

Frequent and chronic pain

- Pain on 54.5% days
- 92% have pain lasting ≥ 6 months

Smith et al, Journal of Opioid Management, 2015
Thompson et al, Pain Medicine, 2014

Socioeconomic impact



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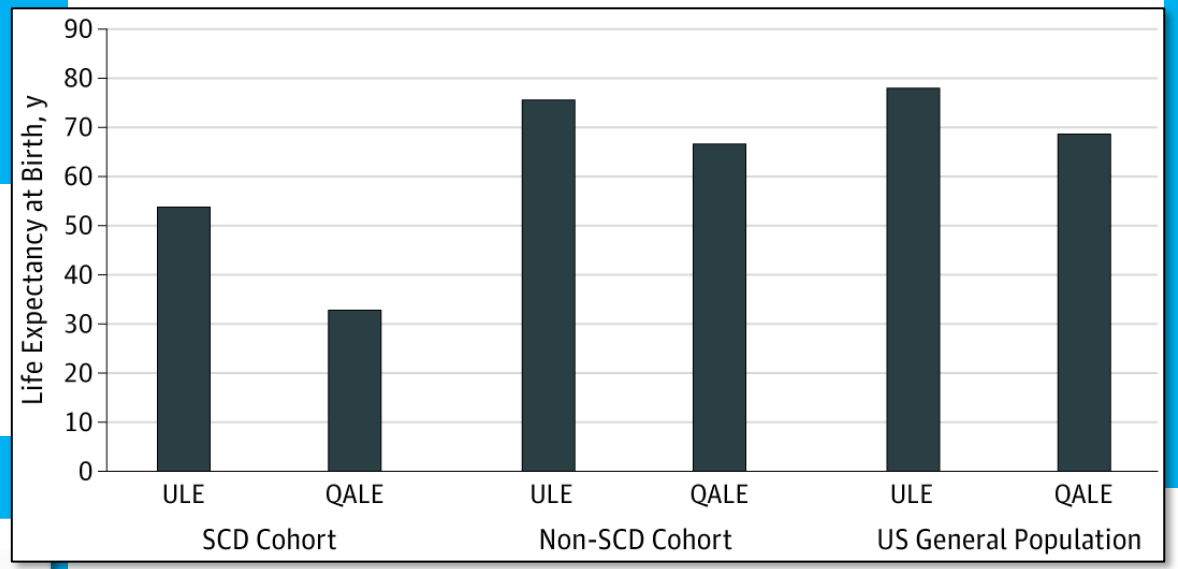

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Limited curative options

Clinical Commissioning Policy: Allogenic Haematopoietic Stem Cell Transplantation for adults with sickle cell disease

First published: December 2019

Prepared by NHS England Specialised Services Clinical Reference Group for Blood and Marrow Transplantation and Haemoglobinopathies

NICE National Institute for Health and Care Excellence

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Exagamglogene autotemcel for treating severe sickle cell disease in people 12 years and over

Technology appraisal guidance | TA1044 | Published: 26 February 2025

Depression and anxiety more prevalent

Treadwell, Lancet Haem, 2023

Thalassaemia: Life Expectancy and Quality of Life Both Reduced

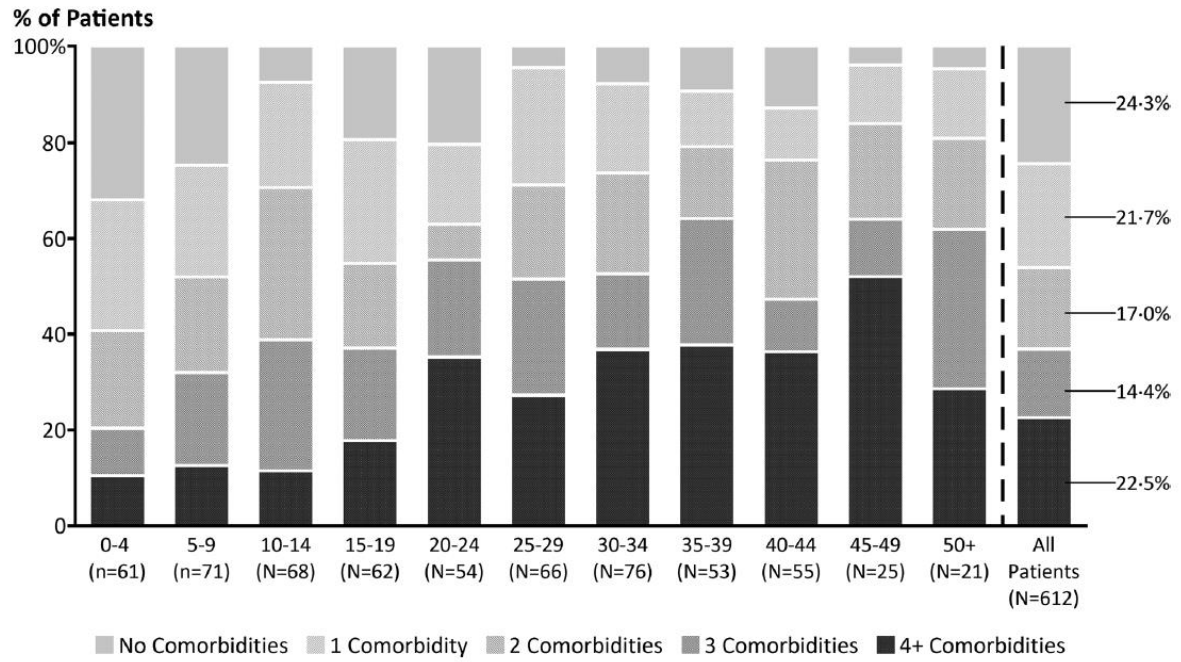


Fig 2. Ten-year (2009–2018) co-morbidity rates by age range in 2009.

Jobanputra, Br J Haem, 2020

Mean age at death
43.9 years

Jobanputra, Br J Haem, 2020

Chronic bone and joint pain in 83.3% including children as young as 3 years of age

UKTS standards 2023

Limited curative options

The NEW ENGLAND JOURNAL of MEDICINE

CURRENT ISSUE SPECIALTIES TOPICS

ORIGINAL ARTICLE

Exagamglogene Autotemcel for Transfusion-Dependent β-Thalassemia

Authors: Franco Locatelli, M.D., Ph.D., Peter Lang, M.D., Donna Wall, M.D., Roland Meisel, M.D., Selim Corbacioglu, M.D., Amanda M. Li, M.D., Josu de la Fuente, M.D., Ph.D., +20, for the CLIMB THAL-111 Study Group* Author Info & Affiliations

Published April 24, 2024 | N Engl J Med 2024;390:1663-1676 | DOI: 10.1056/NEJMoa2309673 | VOL. 390 NO. 18

Strongest Calls for Prevention from People Living with Haemoglobinopathies

‘I can’t believe that thalassaemia babies are still being born in the UK’

49-year-old man with transfusion-dependent beta thalassaemia

3 weekly blood transfusions, history of delayed growth and puberty, hypogonadism, hypothyroidism, impaired glucose tolerance, osteoporosis, chronic bone pain, fatigue not improved by transfusion

‘We need to tell people not to have ‘wrong’ marriages, where they both have sickle trait’

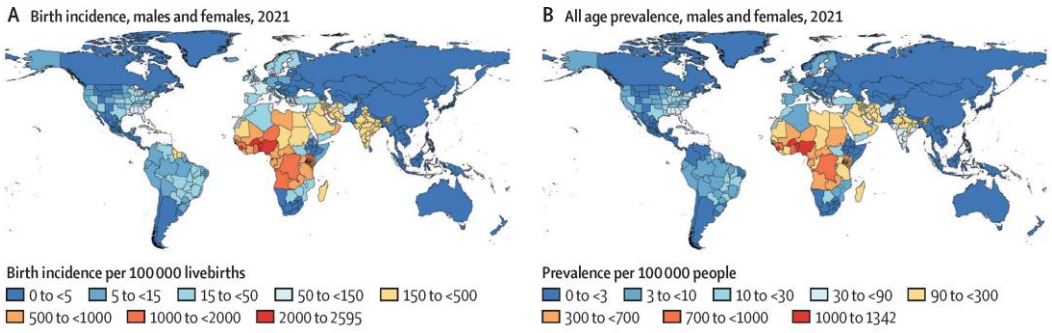
34-year-old woman living with sickle cell disorder HbSS

Frequent severe painful crises, avascular necrosis of the shoulder, severe chronic pain. Regular red cell exchange transfusion programme but severe side-effects of treatment

Increasing Global Birth Rates and Financial Impact on Healthcare Systems

Sickle cell disorder

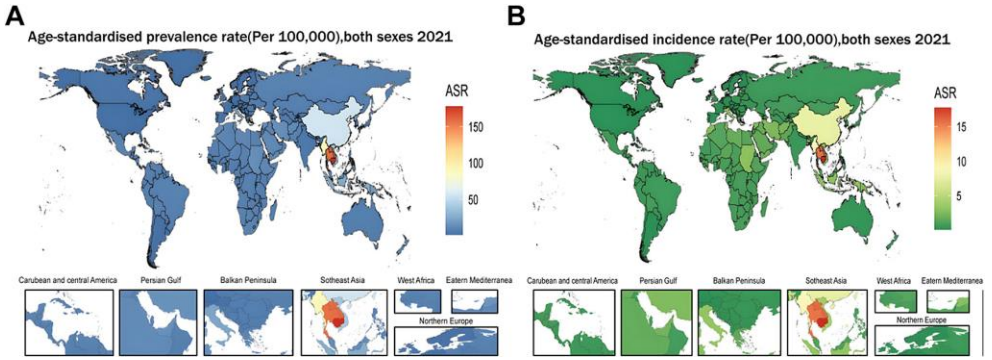
- 7.74 million people globally (2021)
- Increasing number of affected babies born



GBD 2021 Sickle Cell Disease Collaborators

Thalassaemia

- 18.3 per 100,000 persons (2021)
- Slight decrease globally, vast differences between regions



Tuo et al, eClinicalMedicine, 2024

Low & middle income countries: high prevalence, limited resources
High income countries: high cost therapies eg Casgevy® gene therapy & iron chelation, comorbidities

Prevention Approach #1

School-Based Voluntary Screening & Public Awareness

Italy

Latium, Italy

- Voluntary universal screening of secondary school students (13 -14 yrs)
- Health education programme in schools
- Screening by full blood count including red cell indices & HPLC +/- molecular genetics
- Carrier state detected → genetic counselling, family testing
- Started for β thalassaemia prevention, now also sickle cell included
- **1975 - 2011: 1,466,100 students screened; 26,786 (1.8 %) carriers of β thalassaemia identified**
- **Birth rate of affected babies = 0 since 1993** (report 2013)

Amato et al, J Community Genet, 2013

Prevention Approach #2

Pre-marital Testing & Informed Choice

Cyprus

Cyprus: Facilitation of Informed Choice

- Premarital screening for thalassaemia compulsory by law since 1980
- Public education programme
- Premarital population screening & genetic counselling
- Informed, voluntary decision-making : PND > change in partner choice
- **Expected affected birth rate 50 -70 / year → reduced to 0-2 / year in 1980s**

Angastiniotis, Thalss Rep. 2022
Bozkurt, Hemoglobin. 2007
Angastiniotis t al, Hemoglobin. 2021

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Factors for Success

- Societal acceptance including patient advocacy
- Church support
- State funded
- Pregnancies outside marriage uncommon

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Recent Changes

- Confidence in outcomes for affected children
- Migrant populations: different approaches
- Ethical questions around compulsory testing

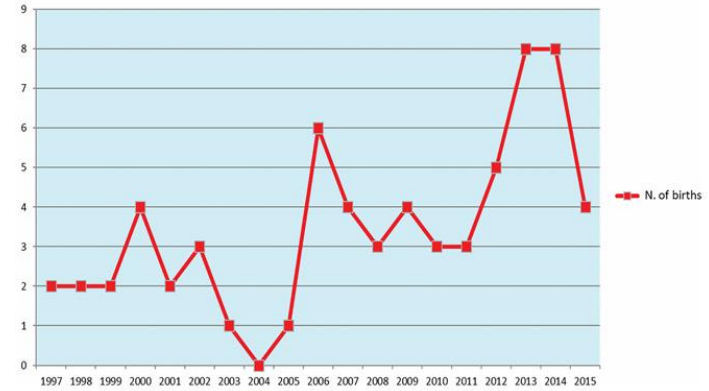

Figure 1. Annual births of babies with β-thal major in Cyprus.

Angastiniotis, Thalss Rep. 2022
Bozkurt, Hemoglobin. 2007
Angastiniotis t al, Hemoglobin. 2021

Prevention Approach #3

Premarital Testing &

Prevention of ‘At Risk’ Marriage

Nigeria

Anambra State, Nigeria: ‘At Risk’ Marriage Forbidden

- Mandatory pre-marital ‘genotype’ of both partners
- Churches and mosques forbid marriage if risk of sickle cell disease inheritance

Medical Genotype Chart Table For Intending Couple

Genotype Partner X	Genotype Partner Y	Possible Combination				Remark
AA	AA	AA	AA	AA	AA	Can Marry
AA	AS	AA	AS	AA	AS	Can Marry
AS	AS	AA	AS	AS	SS	Not to Marry
SS	AA	AS	AS	AS	AS	Can Marry
SS	SS	SS	SS	SS	SS	Not to Marry
AS	SC	SS	AS	AC	SC	Not to Marry
AS	CC	AC	AC	SC	SC	Not to Marry
AA	SC	AS	AC	AS	AC	Can Marry
AA	CC	AC	AC	AC	AC	Can Marry

Note: Three major groups of genotype are: AA (Normal); AS (Carrier), SS (Sickler). Others are SC AND CC.

Anambra State, Nigeria: ‘At Risk’ Marriage Forbidden

- Mandatory pre-marital ‘genotype’ of both partners
- Churches and mosques forbid marriage if risk of sickle cell disease inheritance
- Birth rate & prevalence remain high

Limitations:

- Variable practice across the country
- Lack of universal free testing
- Lack of standardisation in testing, inaccurate results
- Falsified test results
- Pregnancies outside marriage
- Ethical concerns around personal autonomy

Medical Genotype Chart Table For Intending Couple

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SS	AA	AS	AS	AS	AS	Can Marry
SS	SS	SS	SS	SS	SS	Not to Marry
AS	SC	SS	AS	AC	SC	Not to Marry
AS	CC	AC	AC	SC	SC	Not to Marry
AA	SC	AS	AC	AS	AC	Can Marry
AA	CC	AC	AC	AC	AC	Can Marry

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PUNCH

Most Widely Read Newspaper

Love or Lies?: Fake genotype results fuel rise in sickle cell births, broken homes

10th June 2025

Prevention Approach #4

Pre-implantation Genetic Testing

UK

Preimplantation Genetic Testing for Monogenic Disorders (PGT-M)- Approved Conditions

Human Fertilisation & Embryology Authority

- Beta thalassaemia OMIM 141900. *HBB*
- Alpha thalassaemia OMIM 141800; *HBA1*

? includes HbH

- Sickle cell disease OMIM #603903; *HBB (*OMIM 141900)

Preimplantation Genetic Diagnosis (PGD) NHSE Criteria

'PGD represents the only way for parents to have an unaffected child to whom they are both biological parents, without risking the need for termination of pregnancy.'

Eligibility criteria:

- Standard IVF criteria

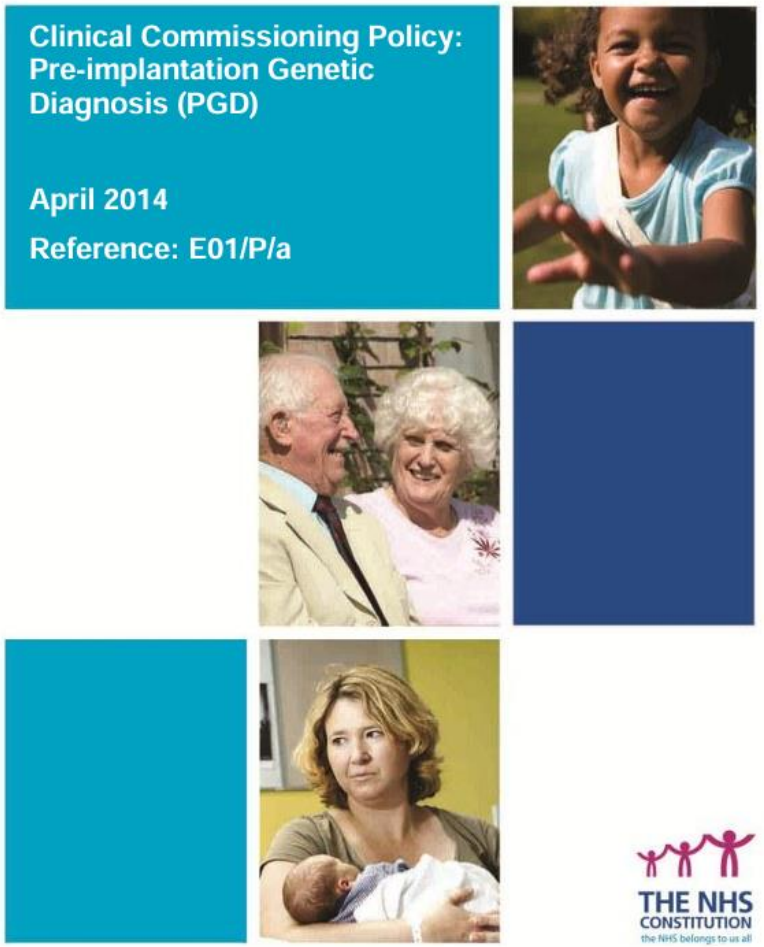
AND

✓At ≥10% risk of having a child with a serious genetic condition

✓HFEA licensed indication for PGD

✓Test included in the [National Genomic Test Directory] (UK Genetic Testing Network)

- No living unaffected child from current relationship



Case Study 1 : 29 year old female with TDT

Patient X (Transfusion dependent thalassaemia)

HBA1/HBA2: αα/αα

HBB: c.47G>A p.(Trp16Ter) – Homozygous

Failed sibling bone marrow transplant in childhood

Partner

HBA1/HBA2: αα/αα

HBB: c.27dup p.(Ser10ValfsTer14) – Heterozygous

Sample ID	Analysis Method	Genotype
	HBA MLPA exon dosage assay (MRC-Holland P140-C1)	αα/αα
	HBA1 DNA sequencing with direct chromatogram check exons 1-3	c.[=];[=]
	HBA2 DNA sequencing with direct chromatogram check exons 1-3	c.[=];[=]
	HBB DNA sequencing with direct chromatogram check exons 1-3	c.[47G>A];[47G>A]
	HBB MLPA exon dosage assay (MRC-Holland P102-D1)	c.[=];[=]

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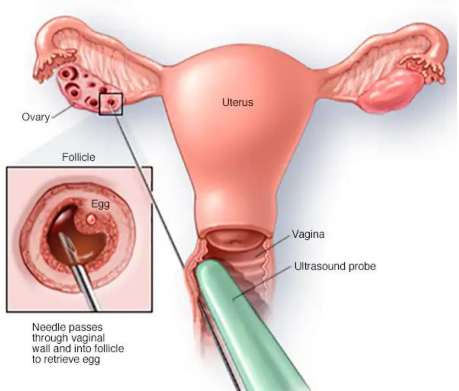
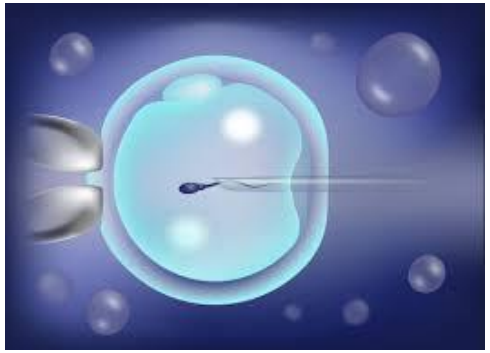
Preimplantation genetic testing

Ovarian stimulation → Oocyte harvest → In vitro fertilisation (IVF) with intracytoplasmic sperm injection → Embryo testing and selection → Implantation and successful pregnancy





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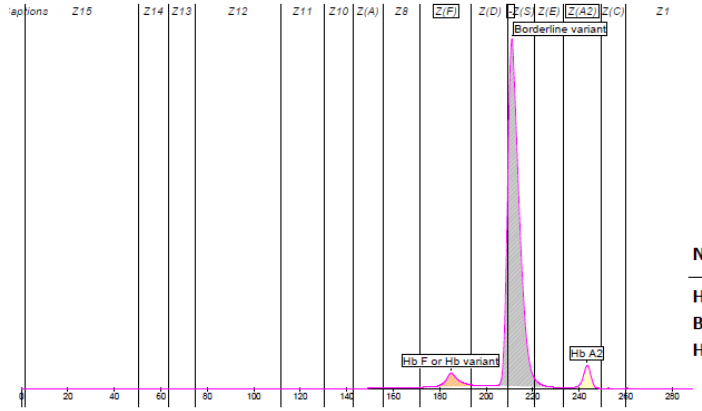
Preimplantation genetic testing

Ovarian stimulation → Oocyte harvest → In vitro fertilisation (IVF) with intracytoplasmic sperm injection → Embryo testing and selection → Implantation and successful pregnancy

Consideration of further PGT cycle 2023 – would be unfunded – decision not to proceed

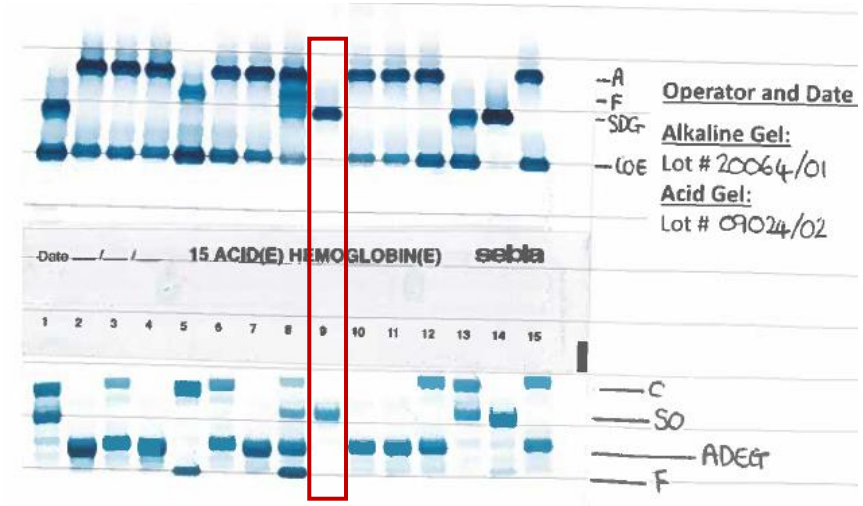
Case Study 2: 33-year old male

Mr Y



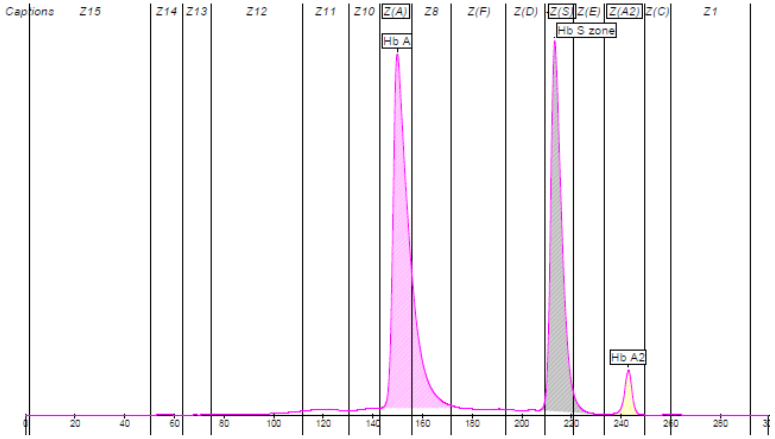
Haemoglobin Electrophoresis

Name	%	Normal Values %
Hb F or Hb variant	3.8	
Borderline variant	92.2	
Hb A2	4.0	



Partner

31 year old female



Haemoglobin Electrophoresis

Name	%	Normal Values %
Hb A	55.6	
Hb S zone	41.0	
Hb A2	3.4	

Case Study 2: 33-year old male

- Mr Y: Hb SS; Partner: Hb AS
- Referred for PGT consideration
- Not eligible due to BMI of female partner: 34 kg/m^2
- Continue to try for natural conception, prenatal diagnosis stated to be unacceptable to couple

Prevention Approach #5

Antenatal Screening & Prenatal Diagnosis

UK

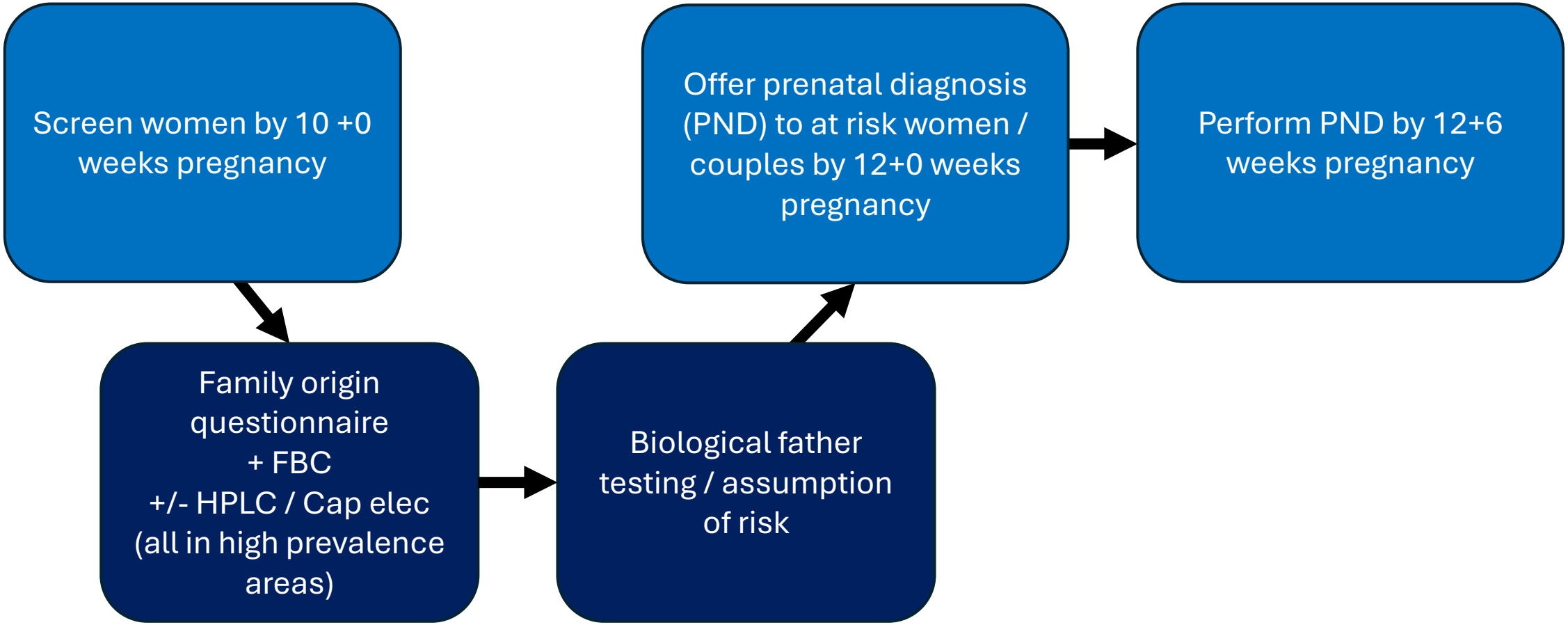
NHSE Haemoglobinopathies Antenatal Screening Programme

GOV·UK

Guidance

Antenatal screening

Updated 10 October 2024



NHSE Haemoglobinopathies Antenatal Screening Programme

GOV·UK

Guidance

Antenatal screening

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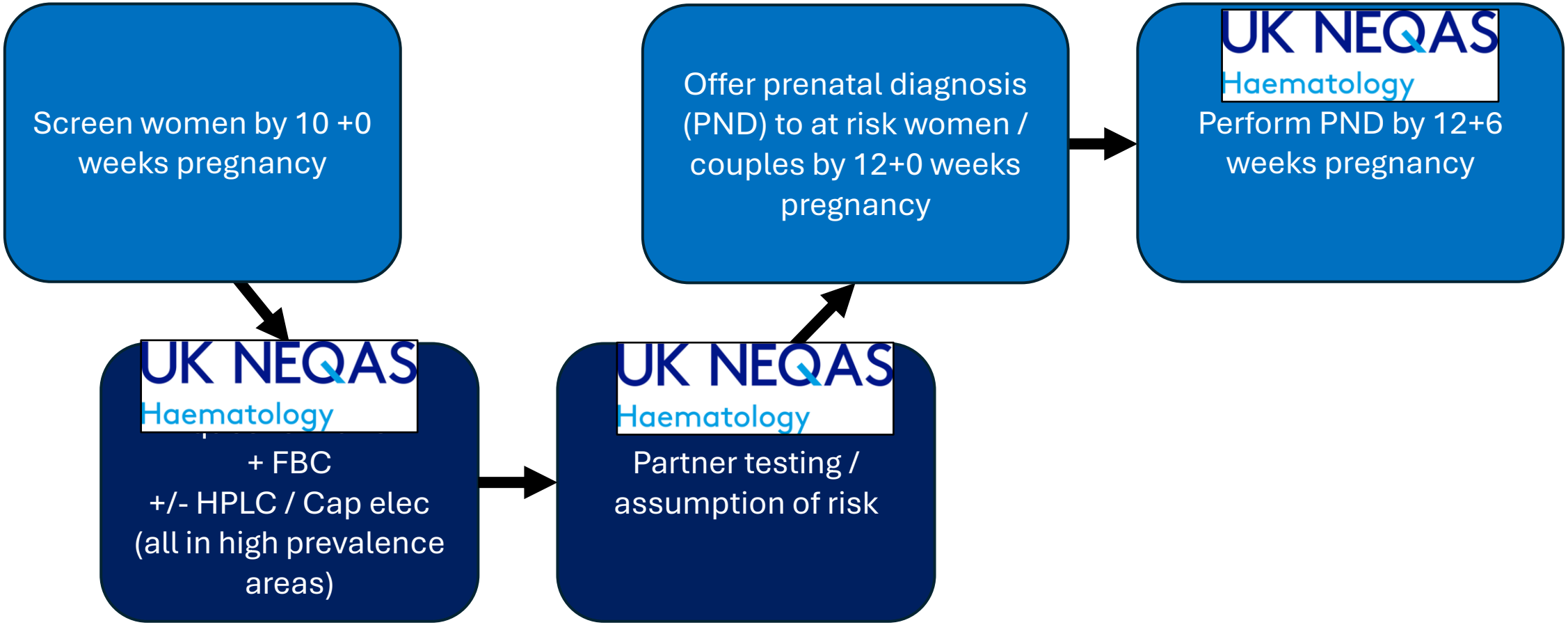

NHSE Antenatal Screening Programme Performance

Office for Health Improvement & Disparities

Corporate report

NHS screening programmes in England: 2020 to 2021

Updated 16 February 2023

Antenatal (AN) screening

99.7%

screening coverage

Measure	Value
Women tested	643,672
Percentage of women tested by 10 weeks	51.3%
Screen positive pregnant women	11,743
Rate of screen positive women	1.92%
Percentage of fathers tested	72.4%
At risk couples detected	802

Prenatal diagnostic (PND) testing

Measure	Value
PNDs performed	399
Affected fetal results	99

NHSE Antenatal Screening Programme Performance

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Prenatal diagnostic (PND) testing

Measure	Value
PNDs performed	399
Affected fetal results	99

Limitations of the NHSE Antenatal Screening Programme

Missed screening: higher risk for women booking late in pregnancy / English is not first language

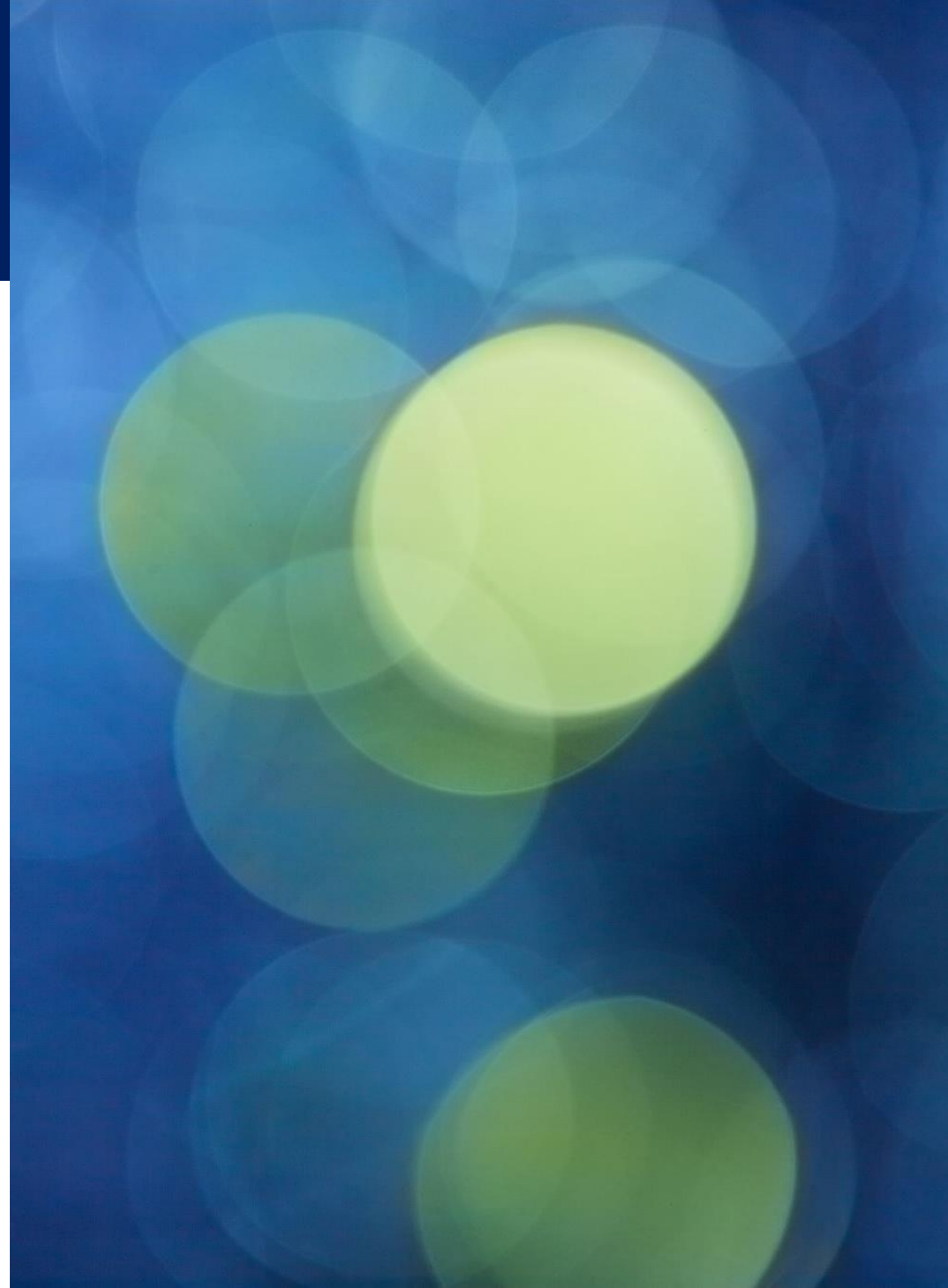
Inaccurate genetic information: family origin, paternity, donor gametes, surrogacy, bone marrow transplant

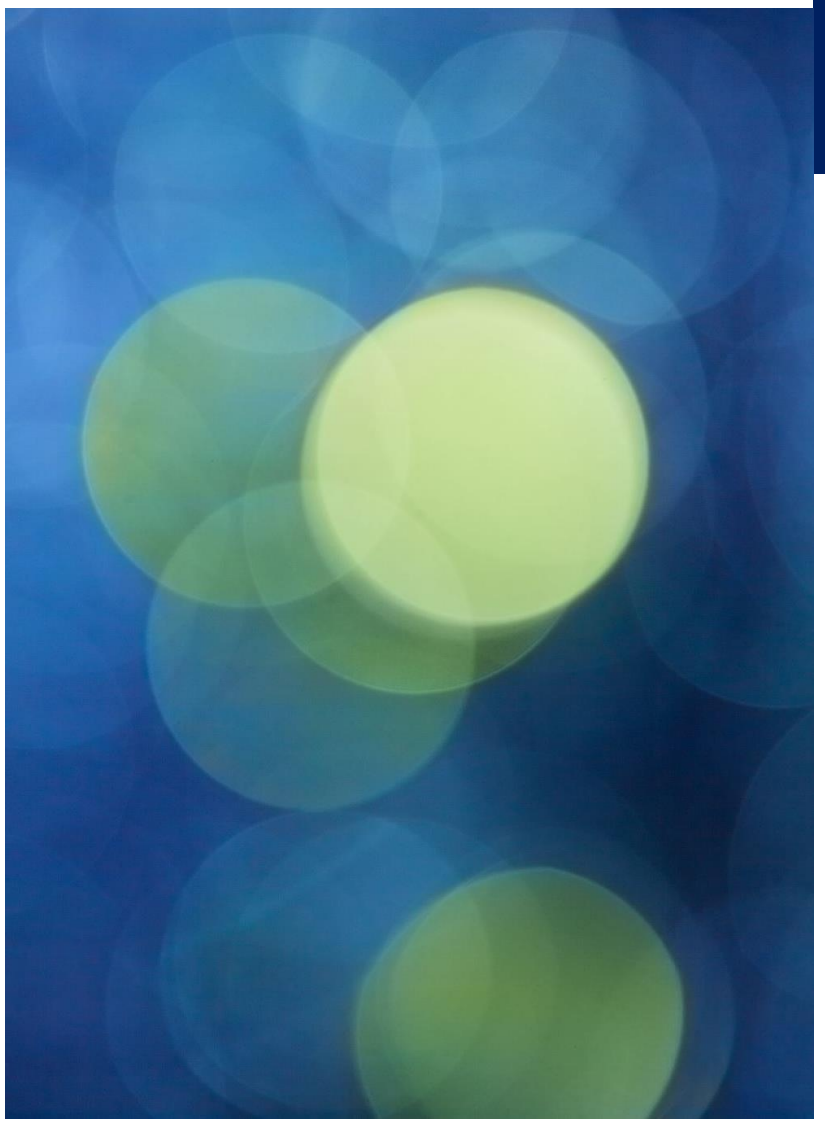
Screening will not detect all at risk couples:

- Restricted testing in low prevalence areas
- α^0 trait outside defined high risk family origins
- Beta thalassaemia trait without raised HbA_2 e.g. severe iron deficiency

Delays / gaps / errors in testing, reporting and counselling

Women who decline screening / PND / termination due to ethical, religious and cultural beliefs





Broadening UK Approach to HBO Prevention

School-based awareness and screening

- Plans to improve access to newborn sickle screening result will miss:
  - ➢People born outside UK
  - ➢Beta or alpha thalassaemia carriers

Consider expanding PGT eligibility

- Need for cost effectiveness data

Avoid sole reliance on antenatal screening & prenatal diagnosis

- Variable acceptance by woman / couple / community

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