Sigdcellesykdom i Norge -Hva vet vi og hva gjør vi?

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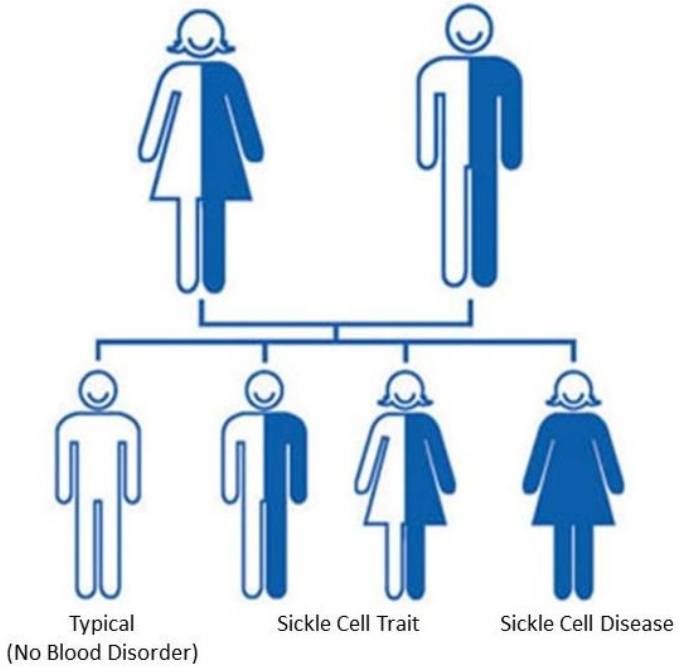
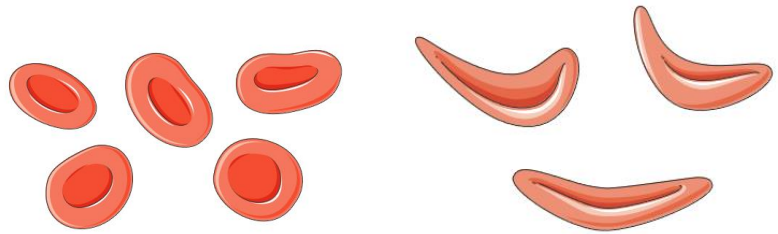
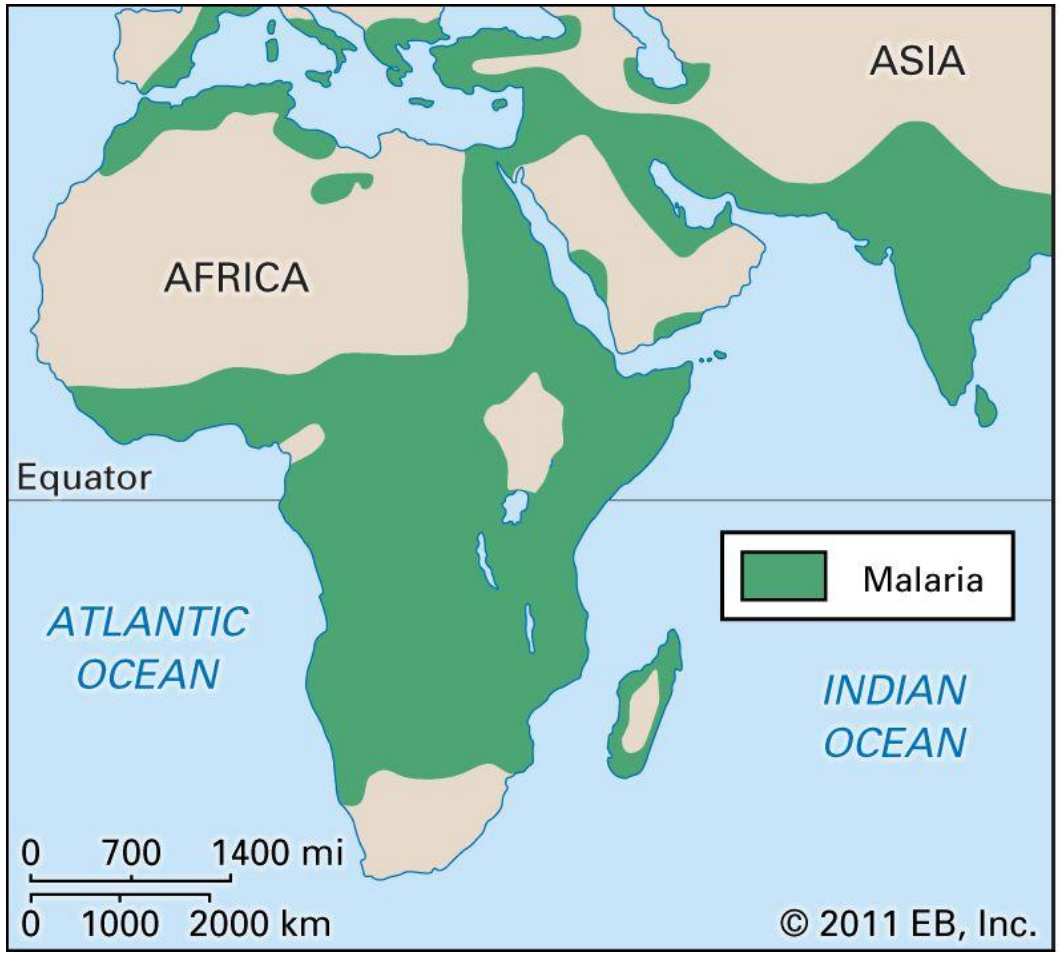
Hva er sigdcellesykdom?

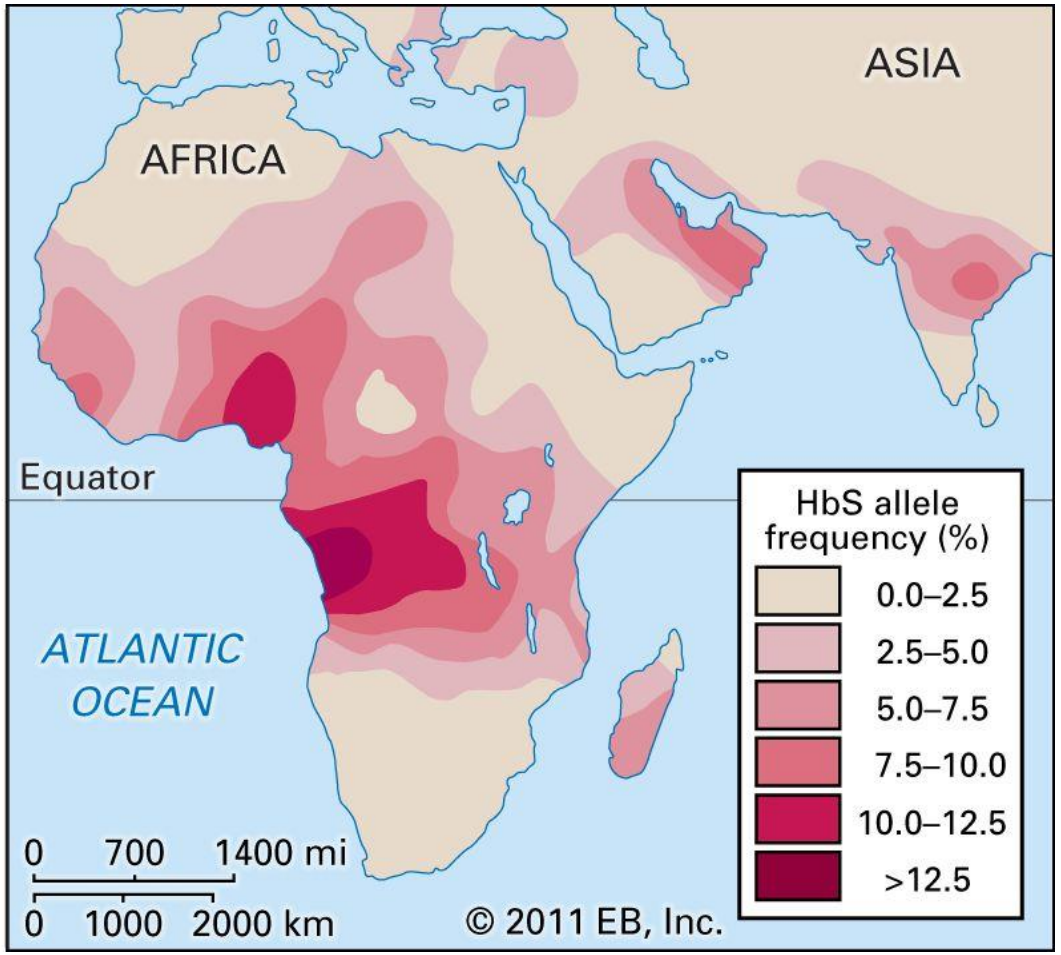
- En arvelig, kronisk sykdom som rammer de røde blodcellene
- Patofysiologi: erytrocyttene sickler, det vil si blir sigdformede
- Bærere av sigdcellegenet har sigdcelleanlegg (sickle cell trait)





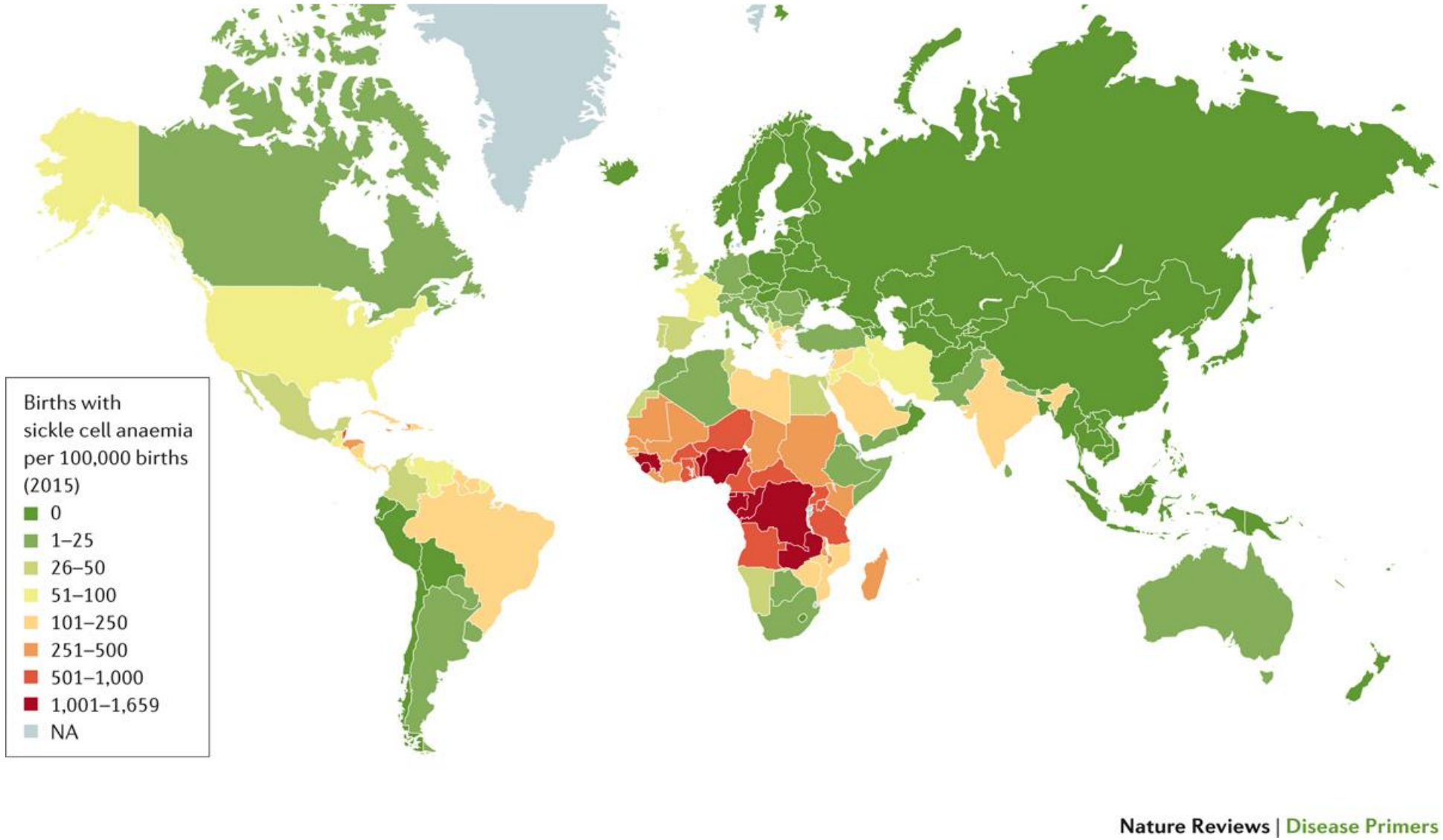
cdc.gov, smart.servier.com





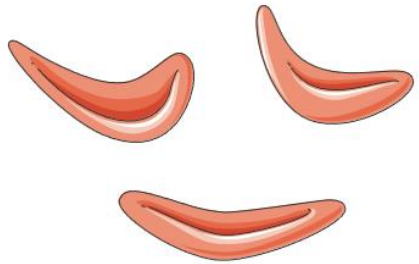
Encyclopædia Britannica, Inc

Map of the estimated numbers of births with sickle cell anaemia



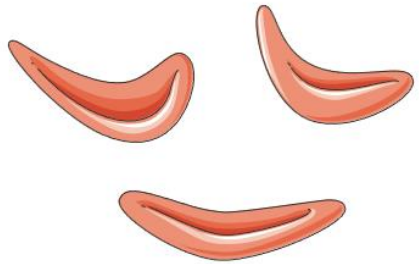
Kato, G. J. et al. (2018) Sickle cell disease
Nat. Rev. Dis. Primers doi:10.1038/nrdp.2018.10

Forekomst



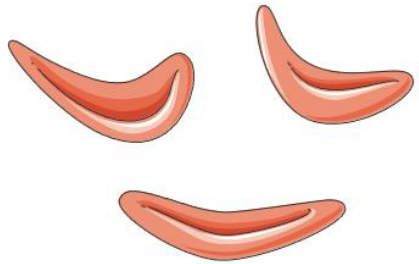
- Ca 300 000 fødes med sykdommen årlig
- Usikker total verdenspopulasjon, men om lag:
  - 100 000 i USA (2010)
  - 67 000 i EU (2020, pre Brexit)
  - 14 000 i England (2017)
  - 10 000 i Frankrike (2021)
  - 3 000 i Tyskland (2017)
  - 3 000 i Italia (2021)
  - 584 i Sverige (2010)
  - 236 i Danmark (2016)
  - 15 i Norge (1997)

Sykdomsmanifestasjon



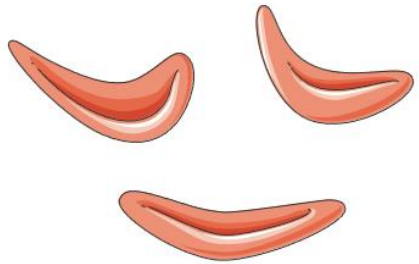
- Anemi
- Vasookklusjon
  - Redusert oksygentilførsel lokalt, kan føre til iskemi og infarkter
  - Kalles vasookklusive kriser eller sigdcellekriser
- Infeksjoner

Sykdomsbyrde



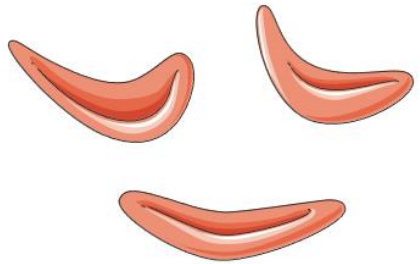
- Økt morbiditet
- Økt mortalitet
- Nedsatt livskvalitet
- Stigma

Hva gir sickling/kriser?



- Nedsatt oksygentilførsel
- Økt oksygenbehov
- Dehydrering
- Å fryse
- Store væromslag
- Stress/påkjenning
- Søvnmangel

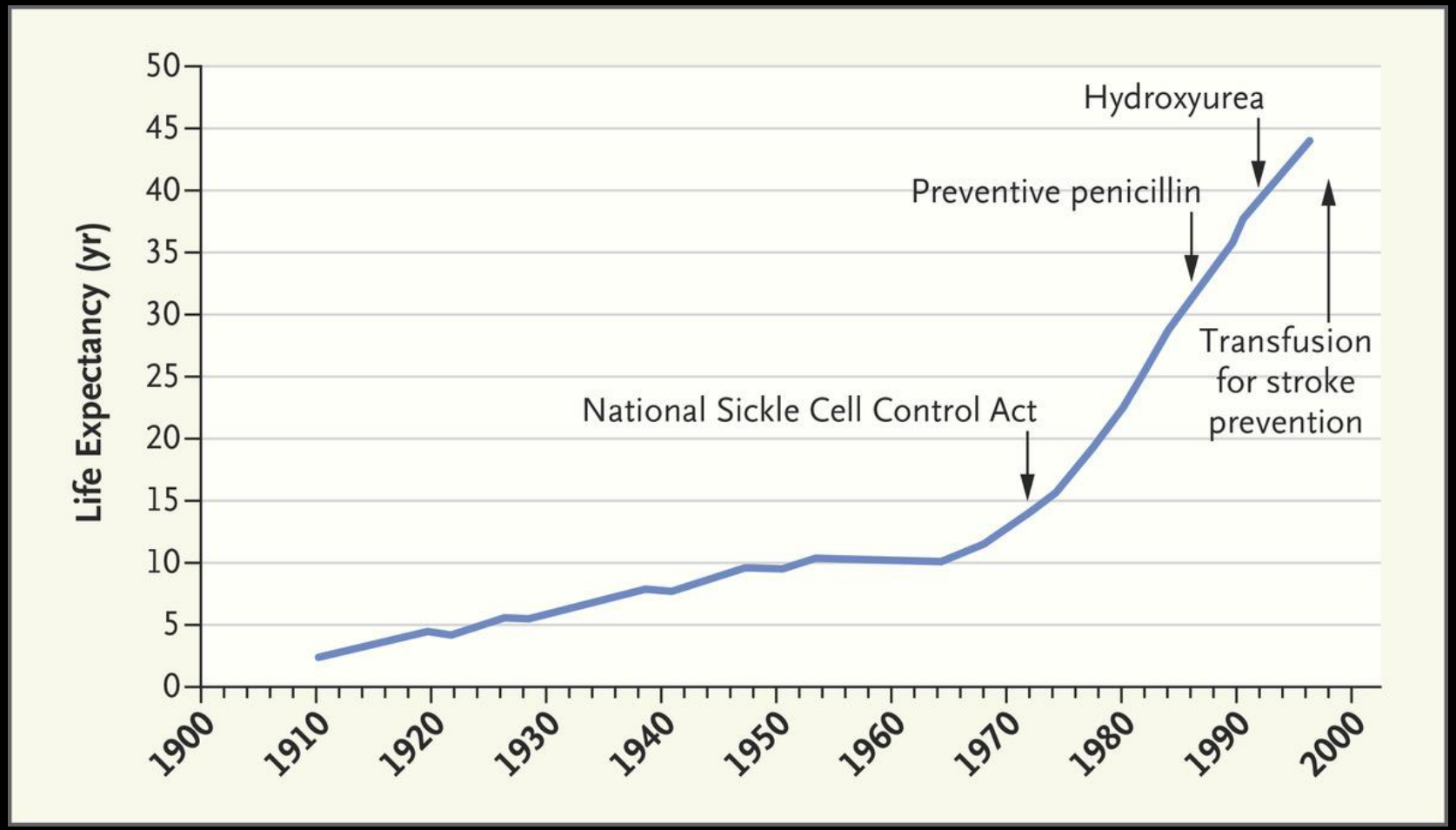
Behandling



- Infeksjonsforebygging (antibiotika, vaksiner)
- Hydroxyurea
- Blodoverføring, utskiftningstransfusjoner
- Stamcelletransplantasjon (kurativt)

- Egenomsorg
  - Unngå å fryse
  - Unngå dehydrering, sørge for rikelig drikke
  - Forsøke å unngå stress
  - Søke helsehjelp raskt ved behov for behandling

Increases in Life Expectancy in Persons with Sickle Cell Disease, 1910–2000.



Wailoo K. N Engl J Med 2017;376:805-807.

The NEW ENGLAND JOURNAL of MEDICINE

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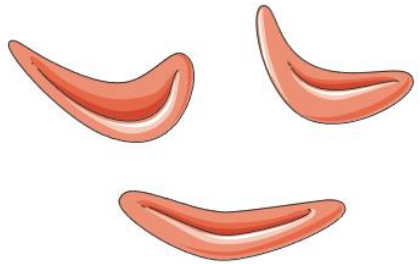


- Lite er kjent
- Økende forekomst
- Enkelte får diagnosen påvist i voksen alder

Newborn screening for sickle cell disease in Europe: recommendations from a Pan-European Consensus Conference

1a. In Europe the burden of Sickle Cell Disease (SCD) has increased and will continue to increase.

1b. It is desirable that all European patients with SCD are enrolled onto registries, with standardized data collection and coordinated follow-up.

2a. The target disease of a NBS programme for haemoglobinopathies is SCD, including all genotypes.

2b. Beta thalassaemia, whilst not a formal target disease of a NBS programme for haemoglobinopathies, should also be reported.

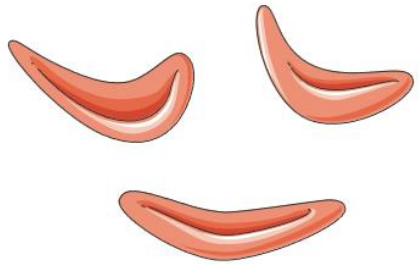
3. Early diagnosis by NBS, together with anti-pneumococcal penicillin prophylaxis and vaccination, coordinated follow-up and parental education, reduces morbidity and mortality from SCD in childhood.

4a. The implementation of a national NBS programme for SCD should be informed by a review of national epidemiological data on SCD, but should not be based solely on a threshold birth prevalence. Where not available, these data should be collected.

4b. A NBS programme should be developed and implemented alongside a national disease management strategy.

Lobitz, Stephan, et al. "Newborn screening for sickle cell disease in Europe: recommendations from a Pan-European Consensus Conference." *British journal of haematology* 183.4 (2018): 648-660.

Likeverdige helsetjenester



- Tidlig identifisering og håndtering av sykdommen (screening)
- Tilknytning til høyspesialisert hematologisk avdeling
- Like godt tjenestetilbud som for andre kroniske sykdommer, tverrfaglig tilnærming
- Like godt tjenestetilbud uavhengig av geografi
- Tilrettelegging i forbindelse med jobb, skole og bosituasjon
- Lærings- og mestringstilbud

Q & A

Informasjon om sigdcellesykdom:

- Innføringsfilm youtube (3 min)
- Center for disease control -learn more about sickle cell
- Sickle Cell Society UK
- handlingsprogram barn
- ASH pocket guides

Sickle Cell Disease (Piel 2017) -med kartoversikt

Sickle Cell Disease (Kato 2018) - med kartoversikt

Artikler om sigdcellesykdom i Europa

- Sickle cell disease as a paradigm of immigration hematology: new challenges for hematologists in Europe | Haematologica
- Immigration and changes in the epidemiology of hemoglobin disorders in Italy : an emerging public health burden | Italian Journal of Pediatrics | Full Text (biomedcentral.com)
- The Impact of Migrations on the Health Services for Rare Diseases in Europe: The Example of Haemoglobin Disorders (hindawi.com)
- IJERPH | Free Full-Text | Genetic Epidemiology and Preventive Healthcare in Multiethnic Societies: The Hemoglobinopathies (mdpi.com)
- One-third of the new paediatric patients with sickle cell disease in The Netherlands are immigrants and do not benefit from neonatal screening | Archives of Disease in Childhood (bmj.com)
- Mortality and causes of death in children with sickle cell disease in the Netherlands, before the introduction of neonatal screening - van der Plas - 2011 - British Journal of Haematology - Wiley Online Library
- Newborn screening for sickle cell disease in Europe: recommendations from a Pan-European Consensus Conference - Lobitz - 2018 - British Journal of Haematology - Wiley Online Library
- Sickle cell screening in Europe: the time has come
- Management-of-Children-With-Sickle-Cell-Disease-in-Europe-Current-Situation-and-Future-Perspectives.pdf (emg-health.com)
- Sickle cell disease in Germany: Results from a national registry (wiley.com)
- Sickle cell disease in Germany: Early insights from a national registry - PubMed (nih.gov)
- Global migration and the changing distribution of sickle haemoglobin: a quantitative study of temporal trends between 1960 and 2000 - ScienceDirect
- Comprehensive care for sickle cell disease immigrant patients: A reproducible model achieving high adherence to minimum standards of care - Colombatti - 2012 - Pediatric Blood & Cancer - Wiley Online Library