Successes and challenges in screening programme for Sickle Cell Anaemia in Africa

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Prevalence of haemoglobinopathies in Central Africa (WHO)
**Environmental and genetics factors**

- **Tropical climate:**
  - malaria endemic,
  - High prevalence of HIV (4-15%)

- **Socio economic conditions:**
  - Poverty: Family income < 5 Usd/day
  - independence since 50 years
  - Few Comprehensive SCA programs

- **Homogenous population**
  - Bantu haplotype: > 80%
  - Alpha-thal deletion: 10-50%
WHO recommended the implementation of national SCA programs in Sub-Saharan Africa region with a focus on widespread awareness-raising, early identification, early access to adequate preventative care, and training of medical professionals.
Limiting factors in SS countries

• Lack of comprehensive SCD program

• Lack of well trained health workers

• Insufficient good and appropriate equipments

• Long delay of transmission of the results

• Difficulties to identify the babies for the confirmation test

• Energy supply most often not available
System of NBS and Early diagnosis

- **Samples**: Dry blood and/or EDTA
- **Emmel Falciformation test**
- **IEF**: basic system (first line method)
- **HPLC**
- **Alcaline Electrophoresis**
- **Solubility test (Itano) in children > 1yr**
- **Capillary Electrophoresis (CE)**
Experience of DRC

• we implemented a systematic newborn and early screening by collecting dried blood spots from newborns and young children aged <5 yrs from maternities and primary health centers in Kinshasa.

A total of 76.991 infants were screened
Model of Dry paper blood collection
## Solubility test (Itano)

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- **Surnageant rouge sans précipitation :** solubilité négatif : **AA**
- **Surnageant rosé avec précipitation :** solubilité positive rosée : **AS**
- **Surnageant clair avec précipitation :** solubilité positive claire : **SS**
Electrophorèse de l’Hb

<table>
<thead>
<tr>
<th>Origin</th>
<th>Carbonic anhydrase</th>
<th>HbC, A₂</th>
<th>HbS</th>
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Confirmation of abnormal Hbs in newborns

Barth’s Hb (alpha-thal)

HbFS in a homozygous
RESULTS

81.5% AA, 16.8% AS, and 1.7% SS. No $\beta^{thal}$ have been found but only few Bart’s Hb suggesting the co-existence of $\alpha$-thal deletion.
Newborn screening programs related to other studies

Most often 1 center or more 2-4 in **capital city**

In some countries centers in different cities

Very rarely in **rural areas**

Prevalence: 1.2% SCA
Program of SCD management in Africa and Madagascar/ AFD funds

En 2006, IIECD (Institut Européen de Coopération et de Développement) et son partenaire congolais CEPCR, s’engagent dans la lutte contre la drépanocytose : ils créent à Kinshasa la Plateforme d’Appui, de Formation et de Veille sur la Drépanocytose (PARCOVED).

Depuis, 100 000 nouveaux nés et jeunes enfants ont été dépistés, contribuant à améliorer significativement leur prise en charge.

En 2014, IIECD lance le Programme d’amélioration de la prise en charge socio-sanitaire des drépanocytaires.
African network: Redac

- Gabon
- Cameroon
- Tanzania
- Centre Africa Republic
- Republic of Congo
- Angola
- Zambia
- Uganda
- Kenya
- Burundi
- Rwanda
- S Sudan
- DR Congo
Interests of Rapid test devices in Africa

- **Cost equipment**: Financial Accessibility
- **Geographical accessibility**: bed to bed; rural areas
- **Reduced delay of diagnosis**: less lost babies
- **Large indications**:
  - Newborn screening (maternities) and early diagnosis (during vaccine schedules)
  - Before any first blood transfusion:
  - Diagnosis of pregnant: introduction of selective screening
  - Prematrimonial test: preventive action
Characteristics of a rapid, point-of-care lateral flow immunoassay for the diagnosis of sickle cell disease

Patrick T. McGann,* Beverly A. Schaefer, Mary Paniagua, Thad A. Howard, and Russell E. Ware

Sickle Scan
HemoTypeSC™: Typical Results

Missing line = positive result
Impact of NBS on Cohorts of SCA patients

Under 5 yrs group increased with the screening program

More data for the natural history of SCD
Early Clinical expressions and late diagnosis
Hand Foot Syndrome (>60%)
Acute anaemia
Splenic Sequestration
Sepsis and pneumoniae (fever)
Bacterial Severe infections (1)

Septicemiae

Pneumonia and meningitis

- High mortality
- Str. Pneumoniae, Haemophilus Infl.
- Salmonella sp
Persistent large spleen (infants >5yrs)

- Abscess
- Aseptic Necrosis
- Malaria?
- Alpha-thal deletion?
- Iron overload
Bone infections: osteomyelitis (2)

- Multiple lesions
- Pathological fract.
- Salmonella sp and staphylococcus sp
TB infections

• Lung infection

• Bone lesions
NTE manifestations

Torrental Nose bleeding:

Hypertrophic tonsillitis
  Upper air obstruction

Glossitis

Dental abnormalities
Leg ulcers

Variable prevalence, but mainly in adolescents and adults

Chronic and often overinfected
Skin lesions: infections, mycosis and/or insect bytes (3)
The burden of SCA

• For the family
  • Family instability
  • Maltraitance
  • Enfant de la rue/ sorcier

• For the society
  • Stygmatization
  • Exclusion
  • Poor schoolarisation

• For the patient
  • Dépression, désespoir
HIGH MORTALITY

• It is estimated that 6. % -9% of deaths in children younger than 5 years are attributable to SCD

• Real rate unknown: misdiagnosis of SCA

• Classical rate: 75% died before 5 yrs of age

• The risk of death in children <5 yrs may grossly underestimate the role of SCD in promoting mortality.
Main Cause of mortality (depending on age of patients)

• **Acute anaemia**: malaria, splenic sequestration

• **Infections**: pneumonia, meningitis, sepsis

• **Acute chest syndrome**

• **Multi organ failure**: renal and cardiac failure
Kinds of available services

• Newborn Screening
  – Gabon – Cameroon - DR Congo
  – Angola – Kenya

• Transcutaneous oxymetry

• Trans Cranial Doppler

• Cardiac echo-doppler : Tricuspidic reflux (PAH)
Therapeutical Approaches

- **Morphin**: not so available (international legislation), fear of this use...
- **Oral Penicillin**: where newborn screening is available.
- **Folic Acid supplementation**
- **Immunization**: high cost, not systematic
  - 7 valent vaccin
  - 23 valent vaccin (Pneumo)
  - 13 valent (available since 2011)
Successes and Challenges
We need new algorythm of NBS and Diagnosis

Thank you/ Merci

Dank U/Aksanti