



Ethics & Societal Implications of Genomic Research on Sickle Cell Disease: An International Perspective

Aceme Nyika, PhD, MPH

Ethics Coordinator

AFRICAN MALARIA NETWORK TRUST

Tanzania Commission for Science and Technology Building
P.O. Box 33207, Dar es Salaam, Tanzania



Outline of presentation

- Introduction
- Issues at the level of the patient
- Issues at the level of the family
- Community-level issues
- Issues at national and global levels
- Concluding remarks



Introduction: Prevalence

- Sickle Cell Disease (SCD) is a global health problem
- However, it mostly affects people of African origin compared with other ethnicities
 - About 90% global births with SCD are in Africa
 - About 1 in 12 African Americans are carriers
 - In Africa, there are 2 deaths compared with 0.13 deaths per 100 patient-years in the USA
- Also prevalent in people of Indian, Mediterranean, Caribbean, etc origins



Introduction: Factors affecting SCD

- SCD is affected by various factors
 - Genetic factors
 - Environmental factors
 - Socio-economic factors
- Interactions of these factors in African settings are bound to be different from the developed countries
 - There is need for empirical research in order to enable evidence-based interventions and policies relevant to African settings



Introduction: Research in Africa

- Rationale for intensifying studies on SCD in Africa
 - Paucity of empirical data on various aspects of the disease from an African perspective
 - Need for application of modern technologies in research on the disease
 - To enhance participation of Africa in efforts to address the high disease burden: collaborative projects to share expertise, resources, etc
- In Africa majority of populations are of African ancestry: less mixed/multiple ethnicities



Introduction: Research in Africa

- Genomic and other types of research on SCD could lead to knowledge critical for development of effective interventions
- Holistic approach including all relevant and complementary specialized areas is needed
 - Health care of patients enrolled in studies: standard of care – treatment, counseling, etc
 - Genotyping, Phenotyping, Bioinformatics, Biorepositories, Technology transfer, etc
 - **Ethical, Legal and Social Issues (ELSI):** as an afterthought....merely to obtain ethical approval



What are some of the ethical and practical issues surrounding genomic research on SCD?



Patients: Poor health care system

- The ideal is to detect SCD before it manifests itself: universal/selective/targeted screening?
- Screening policies are complicated by limited capacity to manage the disease
 - Would it be ethical to have universal screening if health care is not available/accessible?
 - Practicalities of following-up patients/carriers: resources, sustainability after research project,...
 - Majority of women give birth at home (especially in rural areas): some cases may not be captured (could affect data on prevalence)



Patients: Poor health care systems

- Shortages of health care personnel
- Shortages of essential medicines
- Inadequate up-to-date equipments
- Inadequate health care centers
- Poor infrastructure
- Reliance on Traditional Medicine even for health conditions that are better taken care of by 'Western' Medicine (WM)
 - SCD is arguably better handled through WM



Long queues at clinics/hospitals



Patients: Informed consent

- Generally, health care personnel deal with mothers, yet decisions may need both parents (for minor) or family consultations
 - Practical challenges in urban and rural areas
- Language barriers & illiteracy necessitate more efforts to explain technical jargon
 - In terms of resources, time, training of staff, etc
 - Limited vocabulary in vernacular language
- Storage and/or shipment of samples should be explained to sample donors



Carriers: Counseling challenges

- The issue of capacity to provide counseling is critical: carrier vs clinical diagnosis
- If carrier is minor, then it is more complex
 - Counseling of parents initially? Family?
 - Timing of the counseling carrier: When minor reaches adolescence? Challenges of follow-up?
 - Who should be included/targeted then?
- Possible repercussion in light of cultural and or religious backgrounds: beliefs, misconceptions
 - Discrimination/stigmatization in family/community?



The family: Practicalities

- Whereas the principle of autonomy focuses on the individual person, in most African cultures family plays a critical role in decision-making: Immediate/Extended family
- Practical challenges dealing with families
 - Informed consent: before/after testing?; who to include in case of patient/carrier?; etc
 - Counseling: practicalities – at research site or home?; at what point in case of patient/carrier?
 - Beliefs; potential discrimination/stigmatization;



Communities: Acceptability issues

- Culture, religion, socioeconomic, etc affect research & utilization of interventions
 - Prevention, diagnosis, management of SCD
- Challenges in implementing research and interventions: perceptions; fears; beliefs; 'sacred' nature of pregnancy/birth; rumors
 - Dissemination of correct information critical
 - Acceptability of screening: prenatal or neonatal
 - Counseling of patients/carriers/parents in closely knit communities: community engagement critical



National and Global issues

- African countries have to ensure national research agenda includes SCD: Political will
- That would enhance sense of ownership of research and findings
 - could enhance translation of research findings into evidence-based national policies
- Such a participatory approach could ensure sustainability after research projects
 - South-south and South-North collaborative projects facilitate participation of countries



National and Global issues

- International collaborative projects raise complex sovereignty issues such as
 - Specimen & data sharing: shipment and storage
 - Bio-repositories: limited capacity currently
 - Intellectual Property Rights (IPR), Publications
 - Sustainability after the collaborative projects
 - Capacity building
 - Technology transfer, etc
 - Africa to improve national funding of research
- Ethics Committees need to be strengthened



Concluding remarks

- Ethical, Legal and Social issues (ELSI) surrounding SCD in African settings need to be explored empirically and upfront
 - ELSI could be unpacked at the patient/carrier, family, community, national and global levels
 - ELSI could affect effectiveness of interventions
- Community engagement is critical for interventions or policies to be effective eventually



Concluding remarks

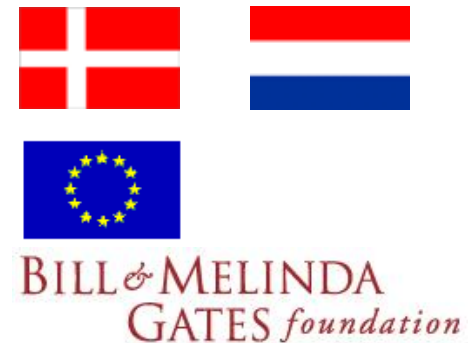
- Collaborative genomic research could go a long way in addressing the problem of SCD
 - pertinent ethical and practical issues should be addressed upfront
- Genomic research should eventually lead to interventions that are accessible to the poor populations that carry the brunt of SCD
 - Should not be for mere academic purposes
 - Collective complementary efforts by stakeholders needed: Public-Private Partnerships could help



Acknowledgements

- AMANET is grateful to:

The Gates Foundation, EDCTP, DANIDA, Dutch Ministry of Foreign Affairs, EC's Directorate General Research, EC's AIDCO,



- AMANET has also received assistance from:

WHO-AFRO, WHO/TDR, AU, SIDA/Sarec, GSK, Wellcome Trust, US-NIH, US-NIMR, US-OHRP



THANK YOU

