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

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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, socioeconomics, 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:
THANK YOU