



Discussion Group A : Diagnostics

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Session 3

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Diagnosics in South Africa

Limited resources

- Only 11 clinical geneticists in SA.
- Geneticists have to trust clinicians that they are educated enough to give genetic counseling and information about the diagnosis. Sometimes clinicians don't have time to read reports
- Informed consent major issue
- Not enough trained professionals for prenatal diagnostics
- Need to educate. There are good act sheets that primary care doctors understand. Idea: Double report to the doctor and the patient to ensure that the information reaches the patient/family
- Limited information about genetic variants in the SA and A population

Whole population newborn screening

Could it could lower costs or increase costs?

What should be screened? Urgent actionable diseases.

Parents started newborn screening in the US

Conclusion: Newborn screening is cost effective and wanted. Many patients with inborn errors live until early or late teens. Burden of state and family

We have to separate screening and diagnostic testing of affected patients

Could diagnostics be harmful/difficult?

- Autism: If diagnosis the child will be excluded from normal schools
- TS a molecular diagnosis can't predict severity
- Families are stigmatized, supernatural causes
- Sometimes good sometimes bad

Discussion of priorities for the future

Need of Education

E.g. access to suitable educational services Discussion of priorities for the future

Patients: Limited access to basic awareness. Many do not even think to go somewhere with their disabled child. Concern from SA in the discussion group: "We are failing to educate about HIV how shall we spread information about complex genetic issues?"

Need to educate people early (school age)

Possible educational pathways: App autism,

Whatsapp: No access to internet but 85% have a Samsung

Clinicians need education about genetics

The need for innovative thinking to diagnose rare diseases in Africa

Ends and starts with the patient and the clinician. Education needed.

Technology goes so fast. Widely available and cheap

Ethics important: e.g autism diagnosis – child has to leave school

Access to all: The middle class can't afford today

Resources, expertise, scientists exist in SA

Need health policy from government

Suggestions:

Digital ICORD courses in rare diseases (FAS, genetics)

Common panels