Greater Sekhukhune-CAPABILITY Outreach Project (GraSCOP) Final Report

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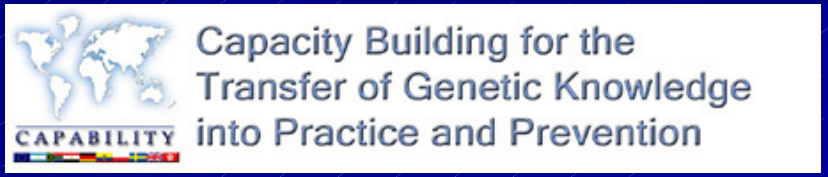


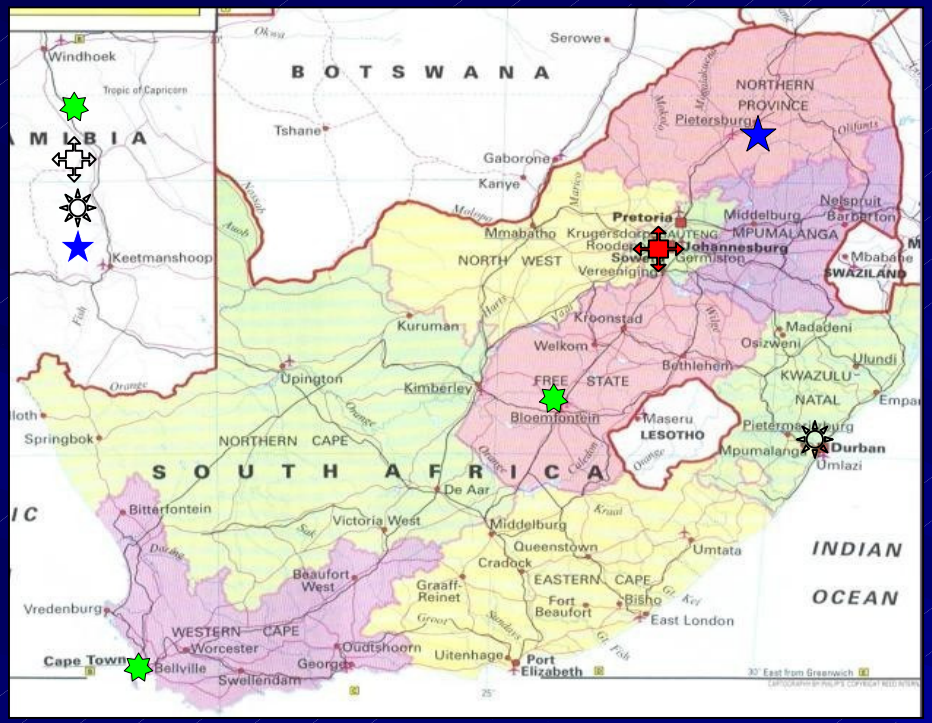
GraSCOP

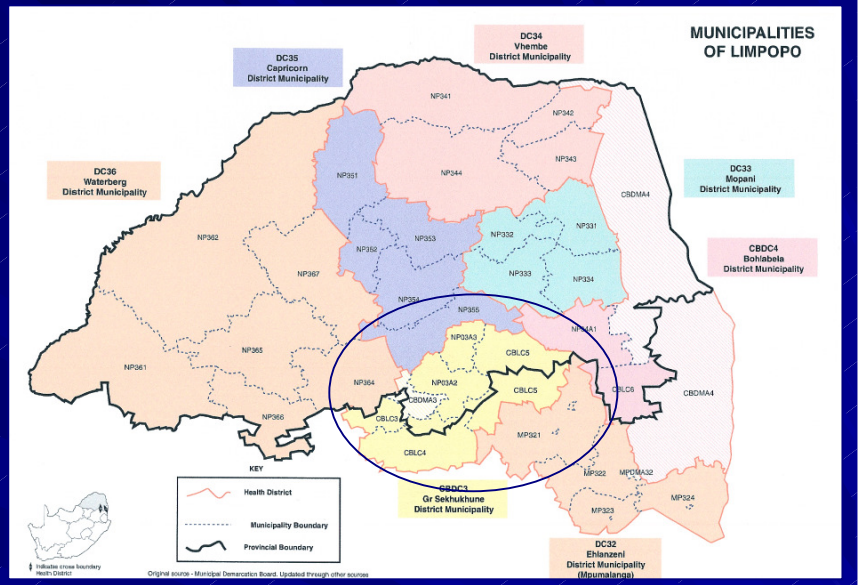
- AIM: to introduce a primary and secondary health care based medical genetic services to Greater Sekhukhune

- OBJECTIVES:

- Test and developing the principles and practices of PHC based medical genetic services outlined in the National DoH's 'National Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities'.
- Further assess & develop the Medical Genetic Education Programme, a national distance learning education programme for post graduate nurse training.
- Evaluate the epidemiology of congenital disorders in Limpopo
- Test the clinical utility of QF-PCR for the postnatal diagnosis of Down syndrome
- Use the knowledge and experience acquired from the project to assist the implementation and development of medical genetic services in Limpopo and other provinces in South Africa.

Greater Sekhukhune





- ~1 million people with ~18 400 annual births
- 1 secondary care & 6 district hospitals
- 65 clinics & 4 community care centres
- 1 paediatrician

Medical Genetics Education Programme Greater Sekhukhune

MGEP Part I

- 44 candidates started the two courses- 39 nurses and 5 doctors
- 6 (14%) candidates DID NOT completed the courses

WHY?

- 28/38 (74%) candidates passed the examination. Similar to previous courses

Medical Genetics Education Programme Tele-teaching MGEP Part I in Limpopo

- Tele-teaching from Pretoria to 4 sites in Limpopo undertaken
- Each site had an on-site nurse previously trained in medical genetics supporting the participants
- These supporting nurses had received training for their role
- 29 nurses received MGEP Part I training
- 25 (86%) passed the exit examination

ClinicalGenetics Outreach Clinics to St Rita's Hospital

- 68 patients consulted over 18 month period

- ALL patients seen were from St Rita's Hospital
- All patients were appropriate referrals
- NO patients were referred from the PHC hospitals attached to St Rita's Hospital

WHY?

WHY?

STAFF VACANCIES

- Doctors 27% (2006)- 35% (2008)
- Nurses 15% (2006)- 44% (2008)
- 41% of all medical professional posts vacant

HIV/AIDS

- 18.5% of population HIV +ve
- 8% have AIDS
- 42% of deaths due to AIDS

TB

- Incidence 173-350/100 000 people between 2002-2007

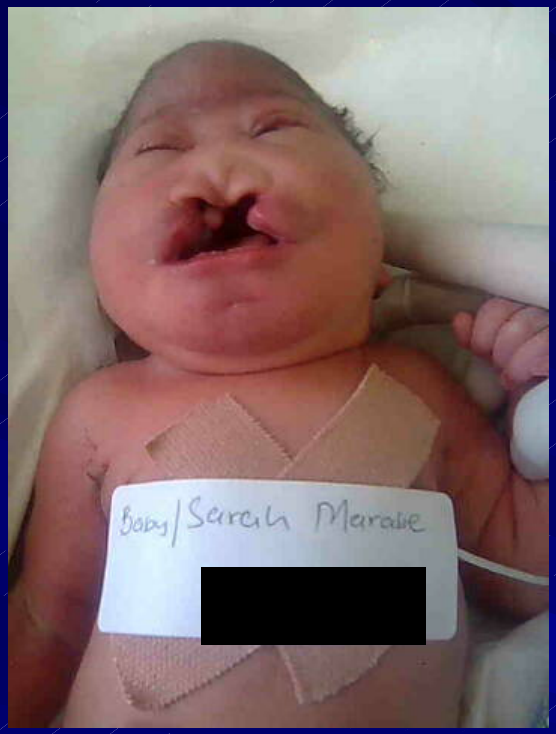
ST RITA'S HOSPITAL

WHY?

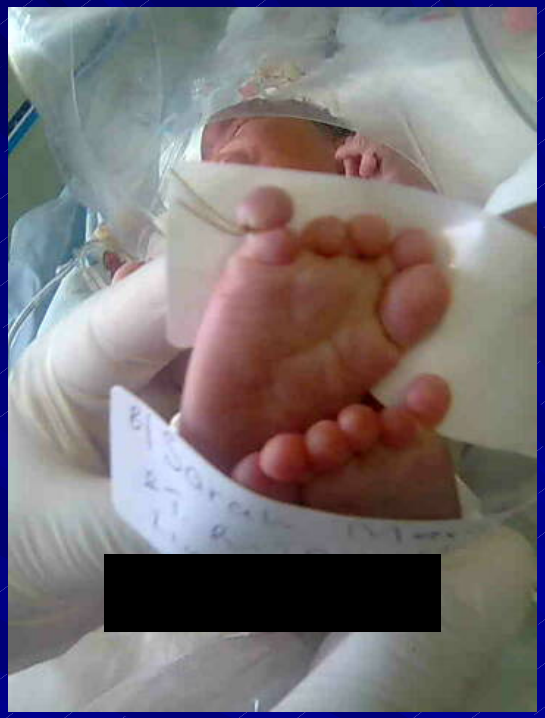
The increasing burden of disease, particularly HIV/AIDS & TB coupled with decreasing staff numbers reduce the significance of congenital disorders.

Available staff just do not have the time to attend to all the problems confronting them and would not be available to be released to attend courses and take patients to St Rita's for the outreach clinics.

GraSCOP birth defect cellphone/photo project



Clinical diagnosis
Trisomy 13
Specimen of skin sent to lab for QF-PCR
Rx. Palliative care
Babe died shortly after birth.
Mother to be counselled at next outreach visit



- Mother's consent obtained
- Clinical photos taken with cell phone & transmitted to Div. of Human Genetics
- Clinical details faxed or discussed during cell phone call.
- Tentative clinical diagnosis, recommended investigations & Rx discussed with medical officer & faxed to hospital.
- Mother & child followed up at outreach clinic.

PROGRAMME WORKING WELL AT PRESENT

Leaves a method of communication with ST Rita's after CAPABILITY

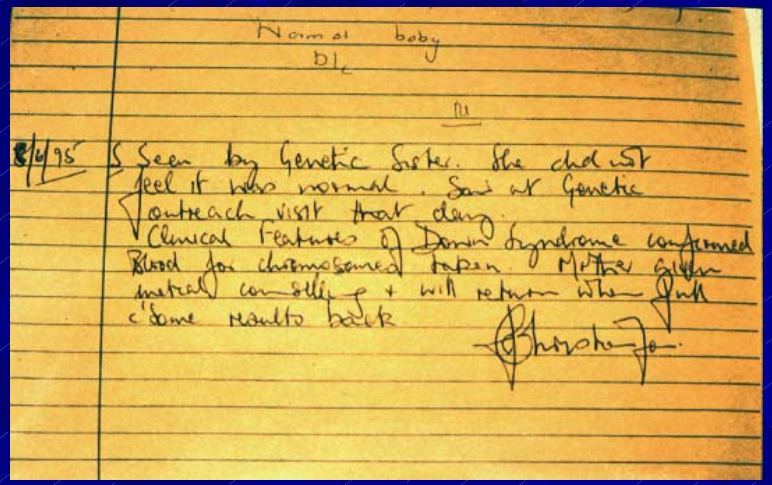
QF-PCR for postnatal diagnosis of Down syndrome- The S African scenario

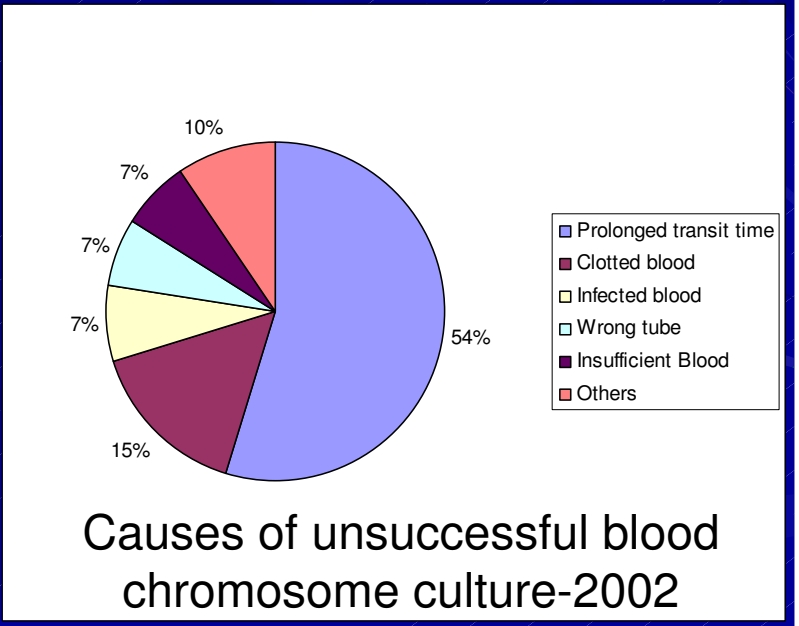
Postnatal clinical practice

- 16% DS infants diagnosed in neonatal period
- <50% DS infants diagnosed before 6 months of age

NHLS Laboratory (Jan 2007-May 2008)

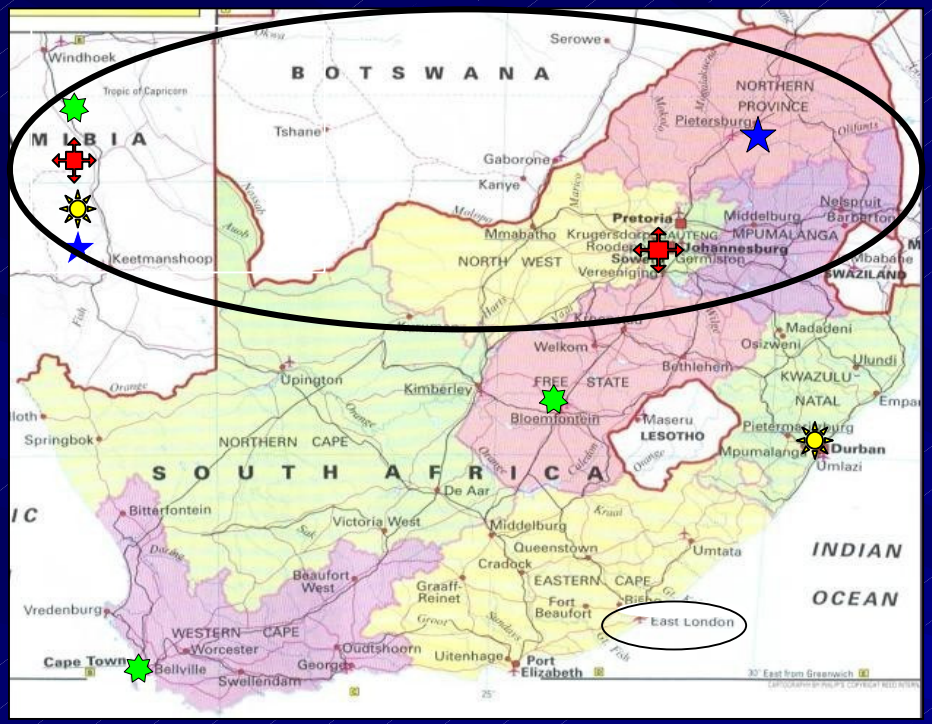
- 653 specimens with DS diagnosis diagnosed with chromosome analysis
- 12% failed culture
- 33% normal chromosomes
- 1% other diagnosis
- 54% DS karyotype
- DS- 95% Trisomy 21
  - 3.6% Translocations
  - 1.4% Mosaics

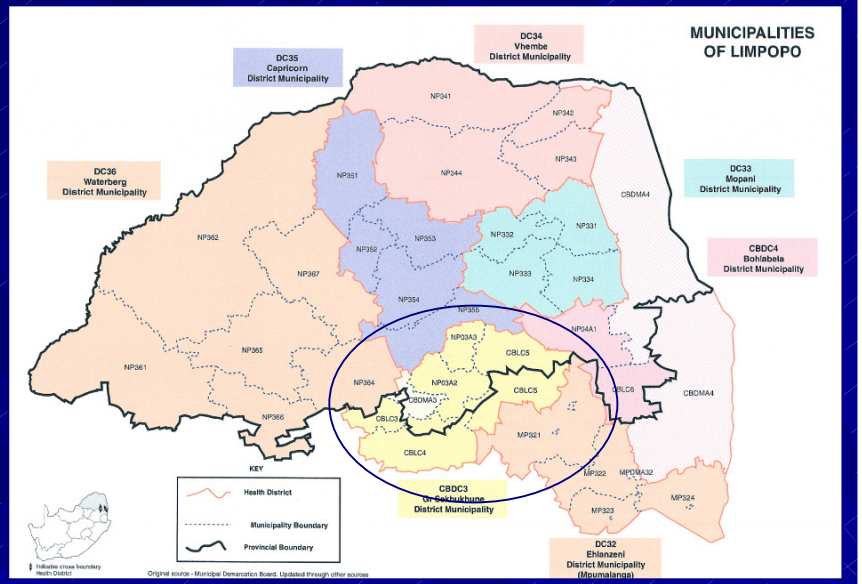




Causes of unsuccessful blood chromosome culture-2002

Clinical utility for QF-PCR for Down syndrome





- Initially the consideration was for QF-PCR for postnatal testing of DS to be undertaken with specimens from Greater Sekhukhune. On request of the paediatricians in Limpopo the use of QF-PCR was extended to the Province
- Because of the staffing crisis in the cytogenetic laboratory all specimens received with a diagnosis of DS are now analyzed by QF-PCR
- The laboratory receives specimens from the 4 northern provinces of South Africa, East London in the Eastern Province, Botswana and Namiba

Cytogenetic laboratory at NHLS & University of the Witwatersrand

2006

- 2439 specimens received
  12 qualified cytogeneticists

2007

- 3193 specimens received
  9 qualified cytogeneticists

2008

- 3227 specimens received
  6 qualified cytogeneticists

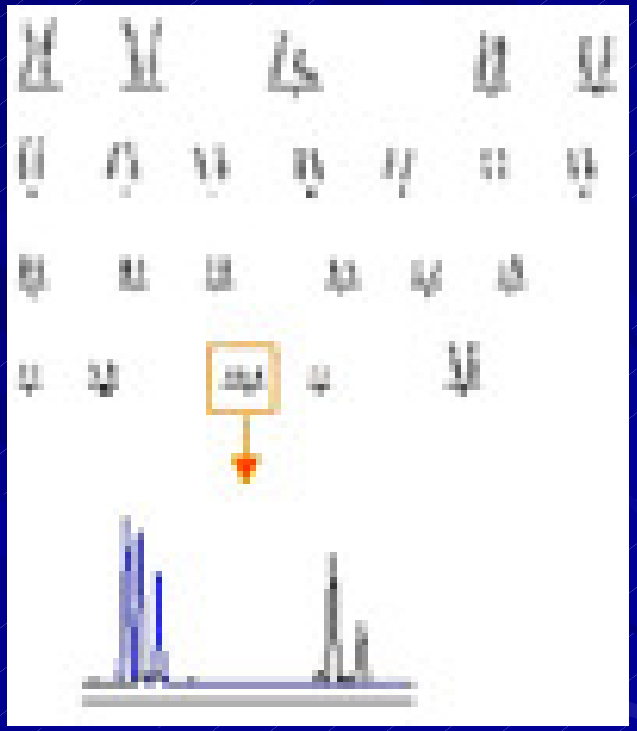
➢ QF-PCR for AMA prenatal diagnosis & postnatal diagnosis of trisomies 13, 18 and Down syndrome initiated

Cytogeneticist- 'a disappearing breed'

Genetic testing for Down syndrome with QF-PCR
July 2008- February 2009

July 2008-February 2009

- 223 requests for DS diagnosis Done by QF-PCR
- 143 (64%) DS diagnoses confirmed by QF-PCR
- 80 (36%) of requests for DS diagnosis not confirmed by QF-PCR



QF-PCR diagnosis of Down syndrome

Suggested EUROGENTEST Evaluation Process for Clinical Utility of Genetic Tests

- ✓ The natural history of the disease if known should be considered so that test & intervention can be properly timed
- ✓ Interventions that might follow a +ve test result should be effective and available
- +/- Qualified pre-test, test and post-test measures including consent processes and genetic counselling, should be in place when needed
- ✓ Financial costs & benefits should be evaluated (30% cheaper)
- ✓ Test services should provide educational material, access to genetic counselling & maintain surveillance over their activities

❖ QF-PCR is now established as the test of choice for the postnatal diagnosis of DS in the Division of Human Genetics

Evaluation of the CAPABILITY demonstration project

+/- Test and developing the principles and practices of PHC based medical genetic services outlined in the National DoH's 'National Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities'.

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Comparison between Chaco & St Rita's outreach programmes

	CHACO	ST RITA'S
Political will & commitment	Good throughout	Present initially
Burden of disease	Decreasing IMR 20$^{0}/_{00}$	Increasing HIV/AIDS & TB
Staffing- numbers	Good Several paediatricians /sanitary area	Poor & getting worse
Staffing- participation	Good /enthusiastic	Limited
Ancillary support	Good Note communications	Poor
Community involvement	Good	Not achievable in present circumstances

Conclusion

In the 1990s a very successful clinical genetics outreach programme was conducted in Limpopo. It formed the basis of the DoH's National Guidelines and many of the principles developed through the programme are accepted internationally.

The inability to establish a similar programme now, because of prevailing circumstances is sobering, & has serious consequences for the future development of medical genetics services in South Africa.

On the 30th October 2009 the DoH held a meeting to discuss the future of these services in South Africa. It was decided to try and obtain funding to undertake a HNA.

This is perhaps the major contribution that CAPABILITY has given S Africa.