



Capacity Building for the
Transfer of Genetic Knowledge
into Practice and Prevention



Demonstration Project Participant 5

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**HEALTH CARE NEEDS ASSESSMENT FOR MEDICAL
GENETIC SERVICES IN GREATER SEKHUKHUNE,
LIMPOPO PROVINCE, SOUTH AFRICA**

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Introduction

Congenital disorders contribute significantly to the burden of infant and childhood disease, death and disability in middle- and low-income nations, including South Africa.¹⁻³ In 2001 South Africa recognised the need for medical genetics services for congenital disorders with the publication of the National Department of Health's 'National policy guidelines for the management and prevention of genetic disorders, birth defects and disability'.⁴ The implementation of these guidelines has been slow, particularly in rural areas, due to competing priorities and limitations of resources.

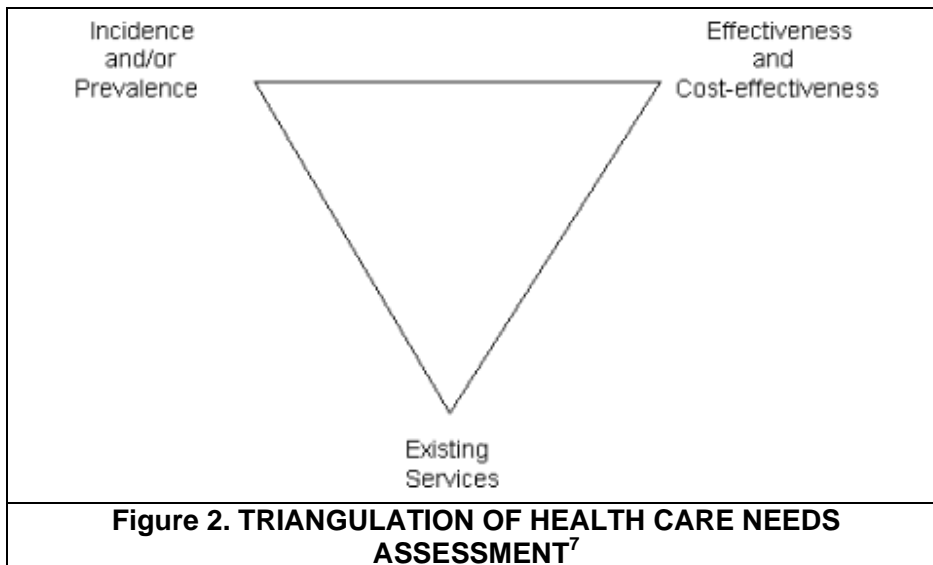
Greater Sekhukhune is one of six health districts in Limpopo, South Africa's northern most province. The health district's southern border lies equidistant from Polokwane (the capital of Limpopo province) and Johannesburg. It is conveniently situated for a collaborative medical genetic outreach programme involving the Limpopo Department of Health, Faculty of Health Sciences, University of Limpopo and Division of Human Genetics, National Health Laboratory Service (NHLS) & Faculty of Health Sciences, University of the Witwatersrand, Johannesburg. It has been chosen for the GREATER SEKHUKHUNE-CAPABILITY PROJECT (GraSCP), a programme to introduce primary and secondary health care based medical genetic services to the district and use the opportunity to:

- test and develop the principles and practices of primary health care based medical genetic services as outlined in the National Department of Health's national policy guidelines and international publications.¹⁻⁴
- further assess and develop the Medical Genetic Education Programme (MGEP), a distance learning education programme currently used by the National Department of Health for post graduate nurse training.
- re-evaluate the epidemiology of congenital disorders in this setting
- test the clinical utility, including cost efficiency, of DNA based medical genetic tests and technologies (QF-PCR, MLPA, Microarray CGH when available) in these circumstances
- Use the knowledge and experience gained from the project to assist the implementation and development of medical genetic services throughout Limpopo and other provinces in South Africa.

Instrumental in the development of the proposal for GraSCP has been the undertaking of a medical genetics service health care needs assessment for Greater Sekhukhune.

Health Care Needs Assessment

Health care needs assessment, through a rational, epidemiological assisted approach, aims to provide information to plan, introduce and beneficially change health care services¹ to improve the health of populations. The main objective of a health care needs assessment is therefore to identify services, activities, opportunities and resources to improve health care. It involves, in the first instance, the assessment of three components referred to as the 'triangulation of health care needs assessment'.⁵ (Figure 1)



The assessment of incidence (birth prevalence for congenital disorders) and (population) prevalence considers the number of people requiring a service or intervention. Effectiveness and cost effectiveness evaluates if a service or intervention confers benefit and the relative cost of the benefit. As health care needs assessment is primarily about change it is necessary to know 'what to change and what to change to'. A baseline of existing services is therefore required to know which services and what opportunities and resources are needed to effect such change.^{5,6}

Demography and Medical Services in Limpopo & Greater Sekhukhune

Limpopo Province (Figure 1) has an area of 123910 sq km (10.2% of the country's landmass) and a population of 5.671 million people, approximately 12% of the country's population. The population is sparsely distributed, approximately 45.5 people per km², and most people (86.7%) live in rural areas.⁷

¹ Health care in this context encompasses diagnosis, treatment, including continuing and palliative care, rehabilitation and prevention.^{7,8} This is comparable to the care and prevention components of a control programme for congenital disorders, which in addition has medical genetic counselling services.^{1,3}

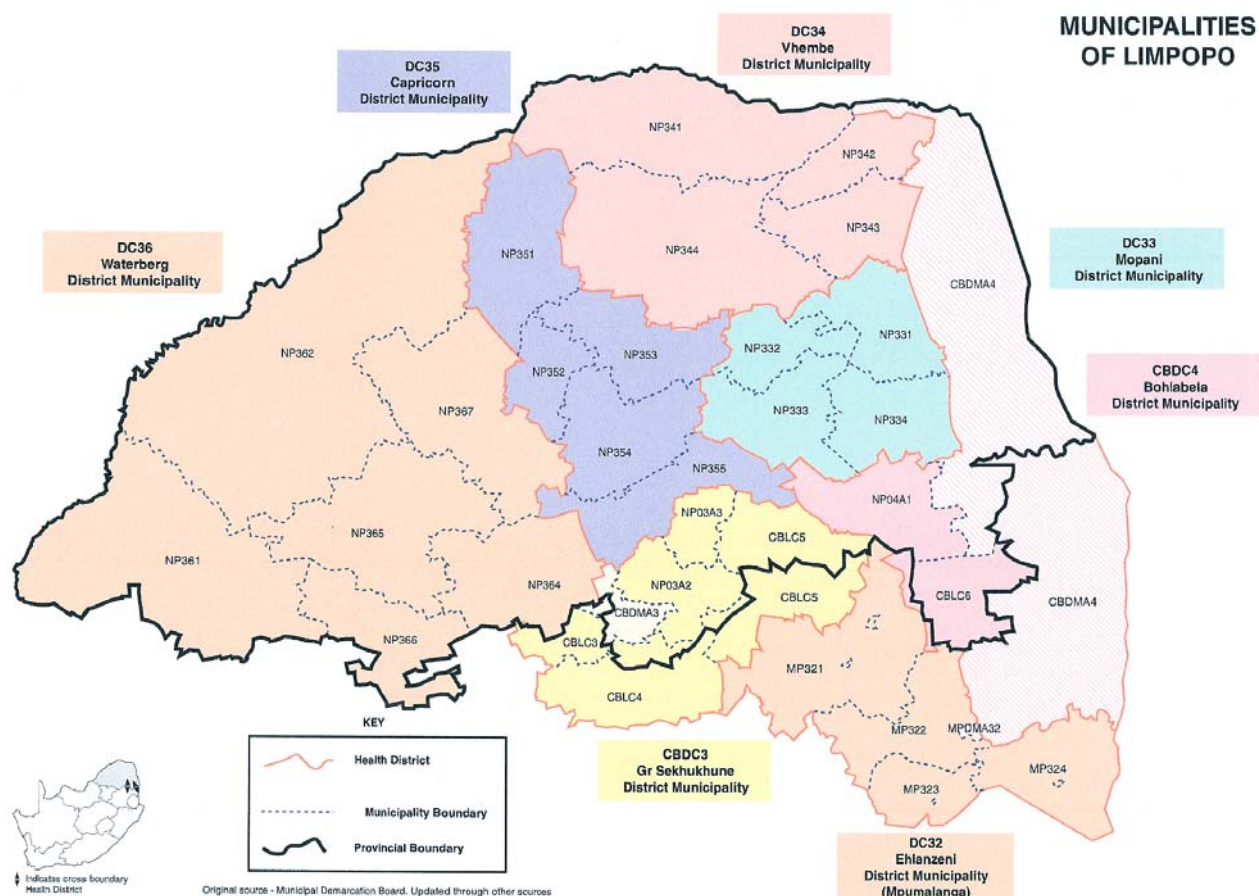


FIGURE 1. LIMPOPO PROVINCE WITH THE GREATER SEKHUKHUNE DISTRICT ON THE SOUTHERN BORDER (YELLOW)

The vast majority (94% or 5.323 million people) of this population depend on the public health care system of 37 hospitals, 30 district (primary care) hospitals, 5 regional (secondary care) hospitals, and 2 tertiary (academic) hospitals, one in Polokwane and the other at Mankweng, 30 km east of Polokwane. These hospitals are supported by a network of 433 clinics, 26 community health centres (CHCs) and 130 mobile clinics (Personal communication-Dr J McCutcheon, Limpopo Dept of National Health and Social Development). The total healthcare budget for Limpopo Province in 2007/2008 was R5429 million (10.6% of national healthcare budget).⁷

The healthcare facilities are staffed by 780 medical doctors, 75 medical specialists and 5827 professional nurses. There are 14.8 doctors/100 000 people and 110.3 professional nurses/ 100 000 people, but 26.8% of medical posts and 15% of professional nursing posts are vacant.⁷

The number of births in Limpopo Province is approximately 140290 annually. Of the women who deliver, 93.4% receive antenatal care and 87.7% of births are assisted by hospital staff and are therefore hospital or clinic based. The Infant Mortality Rate (IMR) in 2007 was 36.2/1000 live births.⁷

Greater Sekhukhune is one of six health districts in Limpopo. It services 1 056 842 people. It has one regional hospital (St Rita's), 6 district hospitals, and primary healthcare is covered by 65 clinics and 4 community care centres. The approximate number of annual births in Greater Sekhukhune in 2005 was 17 000, with 800 births occurring in clinics and 16 200 births in hospitals.⁷

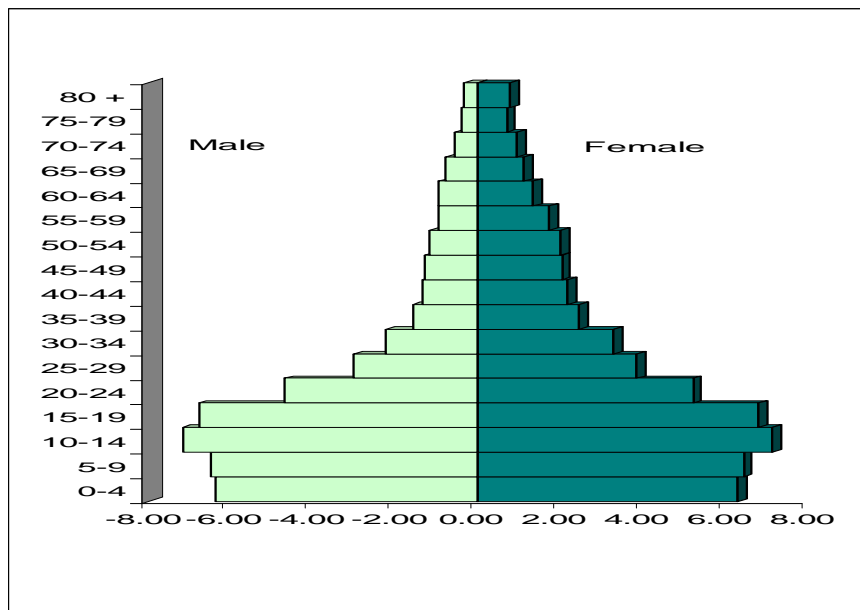


Figure 2: Distribution of population by age and gender in Sekhukhune District

Presently there are no formal dedicated medical genetic services in Limpopo Province including Greater Sekhukhune. Children with congenital disorders, if and when recognised in the district are referred to the Department of Paediatrics at St Rita's Hospital, the regional or secondary care hospital. This Department has Greater Sekhukhune's only paediatrician and three medical officers. Infants and children with congenital disorders who may require more specialised care are referred, if possible, to the Department of Paediatrics, or other appropriate departments, at the Polokwane Academic Hospital. Three times a year clinical geneticists and medical genetic counsellors from the Division of Human Genetics, National Health Laboratory Service (NHLS) and University of the Witwatersrand visit the Department of Paediatrics, Polokwane Academic Hospital.

Medical genetic prenatal screening, diagnosis and associated services are also not available in the province.

Therefore, the integration and development of basic primary and secondary health care based medical genetic services in Greater Sekhukhune, and the linking of these to academic centres in Polokwane and Johannesburg, will add considerably to the care

and prevention of congenital disorders in the district. From there will hopefully extend to the rest of the province.

Epidemiology of Congenital Disorders in Greater Sekhukhune

The estimated annual birth prevalence of congenital disorders of genetic origin in South Africa is 53.4/1000 live births. This is derived from the Modell Birth Defects Database.³ This would translate into approximately 900 infants born each year with a serious congenital disorder of genetic origin in Greater Sekhukhune. No model is yet available to estimate the annual birth prevalence of congenital disorders of teratogenic origin, in developing countries. However, this is considered to be as high as 10-15 % of all congenital disorders.³

Thus 1000 infants (~58.8/1000 live births) with a serious congenital disorder would expect to be born annually in Greater Sekhukhune. Approximately 36 infants with Down syndrome (2.1/1000 live births) and 39 infants with neural tube defects (2.3/1000 live births) would be the two common congenital disorders expected to be born each year.³

In the early 1990s a congenital disorders surveillance programme was undertaken in the rural Limpopo Province, by genetic trained nursing staff doing external examinations only on the first day of life. This documented a birth prevalence of serious congenital disorders of 14.97/1000 live births. High birth prevalence of neural tube defects, 3.55/1000 live births, Down syndrome, 2.09/1000 live births, and oculocutaneous albinism, 0.66/1000 live births were recorded in this study.⁸ Extrapolating from these studies and data from studies in industrialised countries it was estimated that the cumulative birth prevalence of serious congenital disorders by age 5 years in the region may involve 57/1000 live births.⁸ These figures are comparable with those estimated from the Modell Birth Defects Database.³

Mortality of infants and children with Down syndrome was 65 % dead by age two years and 91% mortality for neural tube defects.⁹ It can therefore be expected that the mortality for other serious congenital disorders will be similarly high.

Effectiveness and Cost Effectiveness of Proposed Primary Health Care Based Medical Genetic Services in Greater Sekhukhune

There is limited literature on the effectiveness and cost efficiency of medical genetic services in developing nations. However, it is recognised that that begin to develop medical genetic services once their Infant Mortality Rates (IMR) drop below 40/1000 live births.¹⁰ At this stage of health transition the burden of congenital disorders reaches public health significance.³ The IMR is 36.2/1000 live birth in Limpopo recommending that congenital disorders contribute a significant burden of disease and recommending the initiation of medical genetic services. Discussions with Limpopo Department of Health and Social Development doctors and administrators confirm this.

Given that Greater Sekhukhune has no formal medical services it must be assumed that the implementation of such services, even at a very basic level, will in the first instance improve services for care of infants and children with congenital disorders in the district. At the least it will ensure care- diagnosis, treatment and counselling- for the infants and

children with common congenital disorders, and their parents. For those with more complex problems the service will hopefully lead to early recognition and appropriate transfer for further care. This was the experience during a clinical genetic outreach programme to Limpopo in the early 1990s.¹¹

Cost effectiveness of medical genetic services in such setting has still to be evaluated. Care for infants and children with congenital disorders is expensive and in the long term can affect sustainability of the services. Part of the solution is to ensure the simultaneous development of effective and accessible services for the prevention of congenital disorders.³

However, only audit of the services to be initiated will give an indication of their effectiveness and cost efficiency.

Conclusion

On the basis of this health care needs assessment and past experience a proposal has been put to the Limpopo Department of Health and Social Development to development of primary and secondary health care based medical genetic services in Greater Sekhukhune, one of six health districts in the province. This project, the GREATER SEKHUKHUNE-CAPABILITY PROJECT (GraSCOP), will be a collaboration between the Limpopo Department of Health and Social Development, Faculty of Health Sciences, University of Limpopo, Division of Human Genetics, NHLS and University of the Witwatersrand and hospitals and clinics in Greater Sekhukhune.

Outlined below are the process and details of the project. Negotiations with regional and district health managers are required to ensure that they are fully informed of the project and to acquire their support. The acquisition of middle management's full cooperation is essential for the project to be successful and sustained.¹¹ Discussions with senior medical and nursing staff at the hospitals and in primary health care will also be undertaken to inform them and acquire their cooperation and support.

Education of primary health care practitioners, nursing staff and medical practitioners will form the basis of the introduction of medical genetic services in Greater Sekhukhune. Approximately 50 nurses and medical practitioners from hospitals in the district are being trained with two separate courses of the MGEP (Part 1). The MGEP (Part 1) offers participants basic knowledge on common birth defects and skills necessary for their diagnosis.

The next step will be the establishment of a birth defects surveillance system in the maternity unit of St Rita's Hospital. To support this clinical genetic outreach visits will be undertaken by a team of medical geneticists and genetic counsellors on a monthly basis. At these visits all infants diagnosed, suspected or at risk with a birth defect from the birth defects surveillance system (BDSS) will be reviewed. In addition children diagnosed, suspected or at risk with a birth defect, diagnosed at St Rita's or other hospitals in the district, can be referred and seen at the clinical genetic outreach clinic. In time BDSS and clinical genetic outreach clinics will be established at other hospitals in the district.

In the event of the birth of an infant with a medical genetic emergency, these will be referred appropriately to the Department of Paediatrics, Polokwane Academic Hospital.

The Clinical Unit of the Division of Human Genetics, NHLS and University of the Witwatersrand, will however be available for telephonic consultation when required. In the event of neonatal death the clinical and investigative details will be recorded with clinical photographs and an external post mortem done, where indicated. The case will be reviewed for diagnostic and counselling purposes at a subsequent clinical genetic outreach clinic.

The outreach clinics will also be used as an opportunity to offer in-service training and support to the primary care nursing staff and medical practitioners to improve their clinical and counselling capabilities. With training they can be tasked with taking increasing responsibility for the in-situ care of infants and children birth defects starting with the common ones like Down syndrome, neural tube defects, oculo-cutaneous albinism and clubfoot. This process was pioneered in rural Limpopo Province in the 1990s.¹¹

Data obtained from the BDSS and the clinical genetic outreach clinics will be collected and collated. This will form a database detailing the district's birth defect epidemiology, which can be used to direct the growth and development of the district's medical genetic services.

Further funding will be sought to increase the clinical genetic outreach visits to the district to twice monthly and extend them to other hospitals. Included in this request for funding will be monies to undertake MGEP (Part 2) which includes practical training in clinical genetic diagnosis and medical genetic counselling.

Audit of the programme will be through the patient database that will be developed. This can be compared to expected birth defect prevalence values for serious genetic birth defects. The latter can be estimated as described in March of Dimes Global Report on Birth Defects and using the districts population data.⁵ Further monitoring of the programme can be measured by comparing the accuracy of clinical data on requests for laboratory investigations. Currently, 30% of the specimens with requests for chromosomal confirmation of the diagnosis of Down syndrome in Division of Human Genetics, NHLS and University of the Witwatersrand, have normal chromosomes or some other chromosomal abnormality. It would be hoped that initiating this programme in Greater Sekhukhune will result in a significant improvement in the clinical diagnostic competence of the trained primary care practitioners. Clinical genetic knowledge and skills of trained primary health care practitioners will be assessed before they are trained, in the exit examinations from this training and could be reassessed after one or two years. Finally patient satisfaction surveys, including asking their opinion on how the service can be improved, will be undertaken.

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