

Genetic Services in South Africa

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Practicum Site

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Background

The South Africa Department of Health Division of Human Genetics sought assistance with the development of a questionnaire for the purpose of evaluating and assessing genetic services in the nine provinces of South Africa. The decision to create a questionnaire and conduct an evaluation coincided with the release of the "Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects, and Disabilities" document. The adoption of these guidelines will affect the provision of genetic services in South Africa. Therefore, the Division of Human genetics determined that it is necessary to conduct a comprehensive, systematic evaluation of existing genetic services. Data obtained from this initial evaluation will be compared with data from future evaluations to assess the effect of the newly released guidelines.

Objectives

- Collect information on genetic services in South Africa through Interviews and observations of physicians, nurses, program coordinators, researchers, and public health officials.
- Use information from interviews and observations to develop a questionnaire.
- Pilot the questionnaire and revise as indicated.
- Make recommendations based on interviews, observations, and pilot test of questionnaire.

Timeline and Activities

Weeks 1 - 4: Interviewed and observed physicians, nurses, researchers, program coordinators, and health department officials representing 8 provinces. Developed several drafts of a questionnaire based on information obtained from interviews and observations.

Week 5: Attended the International Conference on Birth Defects and Disabilities in the Developing World.

Week 6: Attended the Southern African Society of Human Genetics (SASHG) Congress.

Questionnaire

The current draft of the questionnaire contains 51 questions divided into six sections:

Part I: Provision of Genetic Services and Availability of Trained Personnel (20 questions)

Part II: Budget Considerations (5 questions)

Part III: Training (9 questions)

Part IV: Genetic Tests (6 questions)

Part V: Data Collection and Reporting (3 questions)

Part VI: Quality of Services (8 questions)

Pilot interviews lasted approximately 1 hour.

Summary and Recommendations

- The provision of genetic services in South Africa varies between and within provinces.
- An evaluation tool that is to be used in all nine provinces must be adaptable to the circumstances and comprehensive in scope.
- Use the existing questionnaire as a starting point and solicit more input from representatives in each province.
- A panel that includes representatives from each province and the national office should review and approve the final questionnaire.
- It is strongly recommended that an evaluation be conducted within the next six months in order to establish a baseline by which to assess the effect of the newly implemented "Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects, and Disabilities".

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