

# Exploring Rare Diseases in South Africa, a Personal Journey: Time for Electronic Record-keeping

## Introduction

To draw robust conclusions about a particular disorder, adequate patient numbers are required. Many hospitals do not keep any records of diagnoses,<sup>[1]</sup> highlighting at least one challenge facing researchers in South Africa. This is on the background of limited funding available to researchers in South Africa, compared with the developed world.

South Africa has a population of more than 50 million people. In the absence of an established National Health Insurance (NHI), there is unequal access to health care. The health-care system consists of a vast public sector and a small but expanding private sector.<sup>[2]</sup> The latter serves 18% of the population, whereas the public health sector serves 82% of the population, predominantly the poor working class indigent and is oversubscribed. The public health sector is challenged by a significant human resource crisis, HIV, and tuberculosis epidemics, and it is also burdened by noncommunicable diseases. Despite the end of apartheid, racial and gender discrimination, income inequalities, and extreme violence continue to impede its function.<sup>[3]</sup> Healthcare varies from the most basic care, offered free of charge by the state, through to highly specialized health services at quaternary level and private sectors for those individuals who are the members of the medical insurance company for those who can afford to pay for it. There has been a steady increase in the number of private hospitals; in 2005, there were 161 and in 2010, there were approximately 238, with 142 of these situated in urban areas.<sup>[4,5]</sup> There are also perceptions among certain strata of the population that the current public health service offers inferior health-care, which lead them to seek healthcare in the private sector, despite the personal financial costs.<sup>[6]</sup>

The mining sector, which provides healthcare for its employees, is responsible for a further 60 hospitals around the country. Secondary care hospitals, or district general hospitals, are found in most of the larger towns throughout South Africa.<sup>[3]</sup> Some secondary care hospitals have computerized data, but many do not keep any records.<sup>[1]</sup> Although tertiary hospitals offer specialized medical care in association with a medical school or university, they do not necessarily have sub-specialist facilities. Five of the nine provinces in South Africa have quaternary hospitals (major teaching hospitals with sub-specialty facilities) with medical schools attached. However, four of the nine provinces do not have sub-specialist clinics for most disciplines. The quaternary hospitals receive referrals from neighboring provinces that do not have quaternary centers.

We were interested in studying Addison's disease (primary hypoadrenalinism) in South Africa, and this serves as an example

to illustrate some of the challenges that we faced in identifying patients with this life-threatening disorder. Since databases in South Africa were not available for Addison's disease, a systematic approach was adopted of initially inviting patients attending quaternary hospitals, followed by patients attending tertiary hospitals and private healthcare facilities. This was followed by inviting prospective participants attending either secondary or primary healthcare facilities.<sup>[7]</sup>

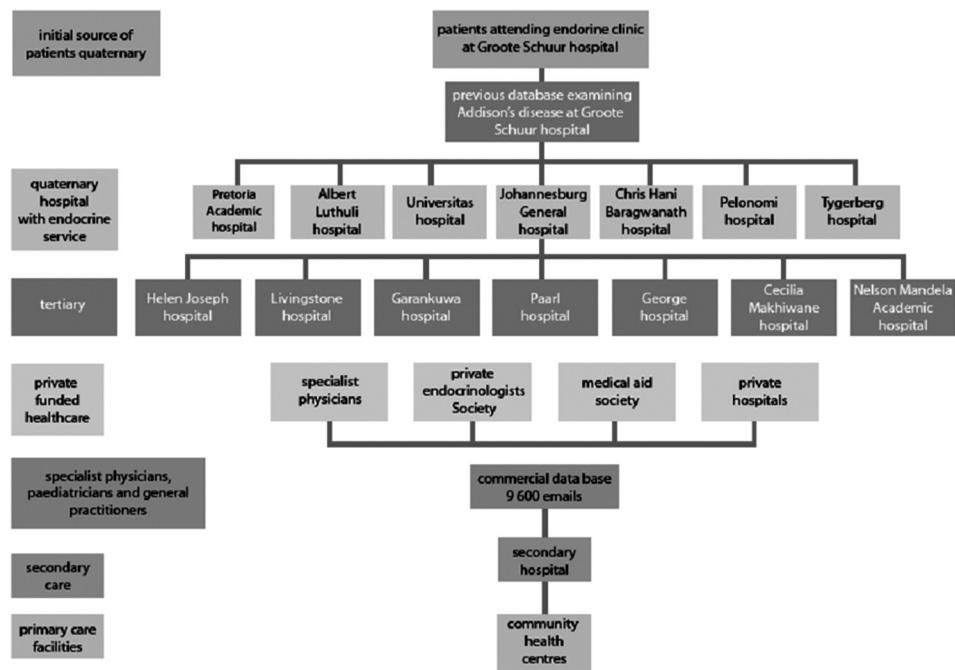
## Method of Registry Compilation

The outline of the procedure followed to identify cases of Addison's disease is shown in Figure 1. As Addison's disease was likely to be diagnosed and managed by endocrinologists rather than generalists, the first phase was to contact all quaternary hospitals to compile a registry of Addison's patients. Patients with Addison's disease who attended the endocrine clinic at Groote Schuur hospital, which is affiliated to the University of Cape Town, were sequentially invited by the medical staff at their routine clinical appointments to participate in this study, as there were no databases to indicate their personal and clinical details.

In the next phase, all endocrinology divisions attached to quaternary hospitals (Pretoria Academic Hospital, Albert Luthuli Hospital, Universitas Hospital, Pelonomi Hospital, Johannesburg General Hospital and Tygerberg Hospital) were contacted to invite their patients with Addison's disease to enroll in this registry. In the subsequent phase, tertiary hospitals without endocrinology divisions were then also contacted to invite patients with Addison's disease to participate in this study. These hospitals included Helen Joseph, Livingstone, Garankuwa, Paarl, George, Cecilia Makiwane, and Nelson Mandela Academic hospitals. All private endocrinologists were contacted to enhance the referral base, using the society of endocrinology membership (Society for Endocrinology, Metabolism, and Diabetes of South Africa) database. All specialist physicians in the Western Cape were accessed using telephone directories, issued by the postal services and letters were written to each of them.

A private commercial database of medical specialists and general practitioners sent 9600 personalized E-mails to all specialist physicians, pediatricians and general practitioners registered with this organization. Specialist physicians or pediatricians were thought more likely to be involved in the management of Addison's disease, compared with other disciplines.

Addison's disease is designated as a medical condition that enjoys the prescribed minimum benefit, and it is a statutory requirement in South Africa that patients belonging to a



**Figure 1:** Systematic outline of the procedure followed or identifying patients with Addison's disease

medical aid have the total cost of the treatment reimbursed.<sup>[8]</sup> Although the medical aid or medical insurance companies were able to access the names of the patients suffering from Addison's disease, only the names of their treating physicians were communicated to the researcher, as divulging the names to third-party would have represented a breach of confidentiality. Thereafter, the treating physician was requested to invite his or her patients to participate in this study.

A facility database of both private and public healthcare was constructed, which included all primary, secondary, tertiary and quaternary healthcare in South Africa. This was created using the Internet and the following keywords "academic hospitals," "clinics," "community health centers," "day hospitals," "Department of Health," "district hospitals," "Hospitals Association of South Africa" and "private medical practitioners", "list of hospitals," "primary care facilities," "private doctors," "private hospitals," "rural doctors association," "secondary care facilities," "secondary hospitals," "secondary tier facilities," "tertiary care facilities," "tertiary tier facilities," were entered into the search engine for each of the nine provinces: "Hospitals."

From each of the websites, the following information was extracted where possible: The name of the hospital facility, the address and telephone number, the name of the superintendent or chief executive officer, the fax number and the name of the liaison officer. Medical practitioners or in the case of medical facilities without doctors, nurses, and nurse practitioners were asked to identify patients with Addison's disease. Letters were written to the superintendents of hospitals, medical practitioners, and specialists, asking them to identify patients with Addison's disease. They were also asked to inform their staff of the Addison's disease national registry.

## Results

There were 35 patients invited to participate at the investigator's center (Groote Schuur Hospital). Three patients from the investigator center declined to participate, citing personal reasons. Letters were sent to nationwide quaternary centers of endocrinology and 44 patients were recruited. No patients were recruited from nationwide tertiary hospitals. Of 9600 personalized E-mails to specialist physicians, general practitioners, and pediatricians, 49 patients were recruited and seven patients were excluded based on an incorrect diagnosis. Of the 110 letters sent to private specialist physicians in the Western Cape, 20 patients were recruited. Moreover, 479 letters were sent to nationwide secondary hospitals and no patients were recruited. No patients were recruited from letters sent to 620 primary care facilities. A total of 148 patients were enrolled in the study. Importantly, we observed that the ethnic make-up of the study population did not match the background population, suggesting some ascertainment bias. The majority of the patients lived in urban areas (87%), and the prevalence was inferred at 3.1/million.<sup>[7]</sup>

## Discussion

Although an extensive effort was made to identify every patient with Addison's disease in South Africa, it is possible that some patients may not have been captured. Collecting the current cohort relied mainly on referrals from quaternary centers and private facilities in the country, where no databases for this disease are kept, except within the domains of the medical insurance companies. Based on data from Løvås and Husebye and Ten *et al.*,<sup>[9,10]</sup> the prevalence of Addison's disease in Western Europe is estimated to vary from 39 to 117 per million, but it has been recorded as high as 144 per million.<sup>[11]</sup> Even using

conservative estimates, this is considerably higher than the 3 per million found in our study. Various factors suggest that this cohort may not have included all of the cases of Addison's disease. Given the demographic profile of the South African population, the majority of participants would be expected to be black, with similar numbers of white and mixed ancestry patients and a small number of Asian patients. Moreover, the vastly lower prevalence identified in this country, compared with Western countries suggests that not all patients were included.

Since Addison's disease occurs even at the highest reported prevalence at fewer than five cases per 10,000 people in the overall population, it may be considered a rare or orphan disease.<sup>[12]</sup> The European Organization for Rare Diseases estimates that there are between 5000 and 8000 distinct rare conditions affecting between 6% and 8% of the population and notes that frequently there are no health policies for these. In addition, there is a paucity of expertise which translates into delayed diagnosis and difficulty in accessing health-care. The benefits of establishing patient organizations for rare diseases is to create awareness among physicians, strengthen cooperation with nongovernmental organizations and to improve the quality of life.<sup>[13]</sup> People with rare diseases often feel marginalized and by researching these conditions, it gives a voice to the disempowered. Anecdotally, our patients responded very positively to participating in this study. They described their gratitude that someone was at last interested in their illness.

Despite the significant burden of communicable and noncommunicable diseases in South Africa, there is still need to research rare diseases. It is particularly relevant that the proposed NHI should accommodate rare disorders and provide a care plan for these conditions.

We acknowledge that the yield from contacting multiple primary, secondary, and tertiary care facilities was very low in the study of Addison's disease in South Africa. On the other hand, the methods employed in this study may represent a blueprint for future researchers undertaking research into unusual conditions, albeit that the methodology may need to be adapted depending on the fields of interest. Robust electronic databases are critical to perform this kind of research, and healthcare providers, especially with the evolution of the NHI should have a vested interest in providing this infrastructure. It is critical that researchers have access to these data so that reliable information reflecting local conditions can be disseminated.

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