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REVIEW

The case for the genetic nurse in South Africa

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Abstract The care and prevention of congenital disorders (CDs) is an emerging but unprioritised health need in South Africa (SA). Inadequate empirical data and underreporting conceal the true burden of CDs while medical genetic services to confront the problem have regressed. Positive epidemiological transition in the country now demands these services are improved to significantly further reduce child mortality. Current sector capacity in SA is inadequate and required personnel targets will not be reached quickly enough to meet the growing health need even if relevant posts are designated. Historically, genetic-trained nurses played a defined role in primary healthcare (PHC) by recognising and diagnosing common CDs and counselling patients and their families, while referring complex matters to the limited tertiary medical genetic services available. Policy changes to redress past inequalities and other healthcare priorities resulted in genetic services being incorporated into PHC, with few genetic nurses retaining their genetic services role. While the medium- to long-term aim for SA would be to develop medical genetic services with appropriate capacity at all levels of healthcare, there is an urgent short-term need to provide basic medical genetic services in PHC. Central to achieving this is the upgrading and re-implementation of the previously successful Medical Genetics Education Programme (MGEP). This post-graduate distance learning, education programme is implemented with the Congenital Disorders Course Book, a distance education tool promoting independent, home-based learning. Together, these tools offer an approach to swiftly build up a nursing workforce with improved knowledge and skills in medical genetics.

Keywords Congenital disorders · Genetic nurses · South Africa · Medical genetics education programme · Education and training

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Introduction

Congenital disorders (CDs) are a common, costly, and critical health issue. Defined as abnormalities of structure or function present from birth, this includes all disorders caused by environmental, genetic, and unknown factors, whether evident at birth or manifesting later in life (WHO 2006). In South Africa (SA), illustrated in Fig. 1, it is estimated that CDs affect 6.8% or one in 15 live births (Malherbe et al. 2015). As for many middle- and low-income countries (MLIC), the true contribution of CDs to the disease burden is significantly underestimated in SA, with national surveillance underreporting CDs by 98% (Lebese et al. 2016). Many remain undiagnosed or are misdiagnosed and the cause of death wrongly attributed (WHO 1999; Christianson and Modell 2004; Nippert et al. 2013). This is largely due to the lack of skilled clinicians to identify and diagnose CDs, combined with inadequate facilities (WHO 1999; Christianson and Modell 2004; Christianson et al. 2006).

As for many MLIC, Millennium Development Goal 4 to reduce child mortality by two thirds by 2015 was not reached

Fig. 1 Map of South Africa showing the nine provinces and neighbouring countries (map by Htonl (own work) [CC BY-SA 3.0 (http://creativecommons.org/licenses/by-sa/3.0) or GFDL (http://www.gnu.org/copyleft/fdl.html)] via Wikimedia Commons from Wikimedia Commons available from https://commons.wikimedia.org/wiki/File%3AMap_of_South_Africa_with_English_labels.svg

in SA. However, rapid reductions were achieved until 2011 (Dorrington et al. 2015; You et al. 2015). Various interventions, including HIV/AIDS programmes and the expanded programme of immunisation, have contributed to bringing the country back into positive epidemiological transition (Kerber et al. 2013; Madhi et al. 2014; Malherbe et al. 2016). As SA develops and communicable diseases are better controlled, the proportion of child deaths and disability resulting from CDs is rising (Alwan and Modell 2003; Malherbe et al. 2015). This follows the trend of high-income countries, where CDs emerged in the 1960s and remain as the leading cause of death in children, accounting for up to 28% of deaths in the under-fives (McKeown 1976; Christianson et al. 2000; Christianson et al. 2006; Malherbe et al. 2015, 2016; WHO 2015).

Despite the lack of empirical data in SA, the previously hidden disease burden of CDs is beginning to emerge through mortality data (Malherbe et al. 2016). In 2013, congenital abnormalities (a sub-set of CDs) overtook infection as the third leading cause of death in early neonates (Pattison and Rhoda 2014). As reported by Malherbe et al. (2016), this trend in early neonatal deaths continued in the Western Cape (WC) in 2014, a province which serves as a healthcare proxy for other provinces in the future (Malherbe et al. 2016).

With the stagnation of the SA infant mortality rate (IMR) and under-five mortality rate (U5MR) since 2011 and neonatal mortality rate since 2009, efforts are underway to further reduce child deaths in SA (Dorrington et al. 2015; Chola et al. 2015; Malherbe et al. 2016). While these interventions will save child lives, none confront the health issue of CDs—limiting the IMR from being significantly further reduced. One example is the 9469 newborn and child lives potentially saved annually by scaling up 11 specific interventions (Chola et al. 2015). These are overshadowed by the 46,754[1] lives that could be saved and/or disability ameliorated every year by implementing appropriate care and prevention for genetically determined CDs alone (Czeizel et al. 1993).[2] Currently, one of the only primary prevention interventions comprehensively implemented countrywide in SA is the fortification of maize meal and wheat flour with folic acid, which has resulted in a 30% reduction in neural tube defects since its introduction in 2003 (Sayed et al. 2008).

CDs have not yet been addressed in SA as a priority healthcare issue in terms of World Health Resolution (WHA) 63.17 of 2010 (World Health Assembly 2010), which outlined specific actions for commitment and allocation of resources by member states. Implementing comprehensive services for the care and prevention of CDs usually begins when a country's IMR is between 40 and 50 deaths per 1000 live births (Modell and Kuliev 1998; Christianson 2000). Despite an IMR of 28 per 1000 live births in 2014 (Dorrington et al. 2015), SA is yet to comprehensively implement genetic services in SA. Such services could be key in significantly reducing child mortality further (Malherbe et al.

[1] Seventy percent of genetically caused CDs based on a prevalence rate of 53.4 per 1000 live births (Christianson et al. 2006) and 1,250,782 live births in 2015 (Statistics South Africa 2015).

[2] Excluding lives affected by teratogens that could be potentially saved, which account for almost 20% of CDs in SA (Malherbe et al. 2015).

2016). While competing health priorities are contributing factors for this lack of prioritisation of CDs, it is now essential that medical genetic services are implemented for this crucial category of non-communicable disease (WHO 1993).

Medical genetic services

Medical genetic services improve health by preventing CDs and reduce suffering by offering care to those affected (Christianson et al. 2006). The key to reducing the contribution of CDs to the burden of disease is to offer the 'best possible patient care in the prevailing circumstances' and prevention so that people affected by or at risk of having children with CDs 'can live and reproduce as normally as possible' (Modell and Kuliev 1998; WHO 1999 2005).

The completion of the Human Genome Project in 2003 highlighted the genetic component of disease, triggering advanced research with many new genetic screening and diagnostic tests becoming available (Secretary's Advisory Committee on Genetics Health and Society 2011). As a result, medical genetics is becoming a field of relevance to the healthcare of many (Guttmacher and Collins 2002). Genetic services are required across the continuum of care but initially focus upon reducing child mortality. As countries transition epidemiologically, the role of genetic services widens to encompass complex multifactorial conditions (of later onset). In SA, the quadruple burden[3] of disease and non-classical epidemiology are impeding CDs from being recognised as significant causes of mortality and morbidity. CDs are the portfolio of the Women's Health and Genetics Directorate under the Women's Maternal and Reproductive Health cluster at the National Department of Health (NDOH). CDs are currently excluded from non-communicable disease (NCD) strategies nationally, negatively impacting their care and prevention.

The lack of capacity

A key barrier to the development of medical genetic services globally is a lack of capacity, impacting industrialised and developing countries alike, albeit on a different scale of magnitude (Secretary's Advisory Committee on Genetics Health and Society 2011). Although inadequate capacity is a widespread constraint in SA throughout healthcare, the impact of these staff shortages in medical genetic services is disproportionately inhibitive on healthcare development given the epidemiology of CDs.

In SA, comprehensive medical genetic services are currently only available at four academic centres,[4] which excludes six of the nine provinces from accessing such services, other than via limited outreach clinics in some areas. Even within the provinces where genetic services are available, access and service vary according to geographical location, and outreach clinics are necessary to penetrate rural areas. Only 11 medical geneticists are practising full-time countrywide (1 per 5 million of the population), and eight genetic counsellors are practising in the state sector (1 per 7 million) (Malherbe et al. 2016) (Shelley McCaulay, Personal Communication, 12 August 2016). Laboratory testing facilities and capacity are also severely compromised.

This capacity falls far short of national recommendations of 27 medical geneticists (1 per 2 million) and 95 genetic counsellors (1 per 580,000) required today to provide a basic universal service (DOH 2003). Until recognition as a primary medical specialty in 2007, medical genetics was a sub-specialty under which many registered through a grandfather clause in 1999 (Kromberg et al. 2013). Although 17 medical geneticists have qualified since 2001 and six registrars are currently in training, this additional capacity has been offset by a loss of 19 to the sector. Seven have retired, six have emigrated, two have died, two moved to private practice, and two are not currently practicing. Genetic counsellor numbers are similarly limited with many of those qualifying remaining unavailable to public service as posts have been closed or frozen, forcing their emigration or to move to other fields or the private sector, where seven are currently practising.

With limited posts available and few doctors choosing to specialise in medical genetics within the greater context of a doctor shortage in SA,[5] it is unlikely that capacity targets will be reached in the medium term. With only four training centres available to train medical geneticists countrywide[6] and only two centres[7] training genetic counsellors and a severe shortage of allocated posts, these circumstances necessitate other options to be considered for the more immediate expansion of services. This speaks to the role of allied healthcare professions, specifically nurses, who can undertake a key supplementary role in genetic services.

[3] The quadruple burden of disease in SA includes HIV/AIDS and TB, violence and injuries, high maternal and child mortality, and non-communicable diseases.

[4] Comprehensive genetic services are available at the University of Cape Town, University of the Free State, University of Stellenbosch, and University of the Witwatersrand.

[5] Sixty doctors per 100,000/population in 2013 compared to the global average of 152/100000 (ECONEX 2015).

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History of genetic nurses in South Africa

While the potential role of nurses in genetic services is not new, their impact continues to be largely unappreciated. Appropriately trained nurses can provide an initial filter for referrals to the geneticist and perform an educational role (Emery and Hayflick 2001). By diagnosing, counselling, and treating common CDs and recognising and referring more complex disorders as necessary, genetically trained nurses provide a major contribution to genetic services (Christianson et al. 2000; Ehlers 2002). In low resource settings in SA, especially rural areas, nurses in PHC contribute significantly to antenatal, labour and delivery, and newborn care. They live and serve locally, understand the local language and culture, and are well respected in the community, making them ideal candidates to be trained as point of care genetic nurses and genetic nurse counsellors (Alwan and Modell 1997; Christianson et al. 2000; Christianson et al. 2006).

Nurses were identified as a key component of medical genetic services early on in SA. The first genetic nurse was appointed in Durban in 1974 with the mandate to 'find cases, follow-up affected families, create general awareness of genetic services, and coordinate existing facilities' (Op't Hof and Roux 1983). By 1977, a network of 16 genetic nurses countrywide had developed and were linked with medical schools, provincial services, and their existing clinics and diagnostic laboratories, based around major urban centres (Op't Hof and Roux 1983). Genetic nurses were senior nursing personnel who underwent intensive training to effectively deal with and counsel patients with common disorders. During the late 1970s, the PHC nurse cadre was established enabling nurses with the training and authority to assess and diagnose patients, prescribe treatment, and dispense medication (Kautzkyi and Tollmani 2008). Beyond the role of nursing counterparts in high-income countries, this was necessity for countries such as SA due to the lack of medical practitioners. Ad hoc training of genetic nurses spanning a few days to several weeks continued into the 1980s (Kromberg et al. 2013).

These genetic services mainly benefited the middle class, white population in urban areas. In 1985, only 18% of the 4856 patients seen at genetics clinics were black South Africans, despite making up 74% of the country's population (Jenkins 1990; Christianson et al. 2000). Efforts were made in 1990 to expand into more rural areas with no genetics services but were prevented by budgetary constraints (Jenkins 1990).

Collectively, all these factors resulted in genetic nurses becoming the 'back bone' of the genetics service, often working in extremely challenging conditions without medically qualified supervisors, with only five medical geneticists in the country at the time (Jenkins 1990).

The Northern Province experience

The shortfall in medical genetic specialists in SA necessitated outreach programmes to take this expertise where it was lacking. One of the best documented programmes was a clinical genetic outreach in rural Limpopo (then Northern Province) (Christianson 2000; Christianson et al. 2000). Initiated in 1989, this collaborative project[8] spanned 7 years (1989–1996) and reached an estimated fifth of the population of the province (Christianson et al. 2000). By 1992, week-long clinics were held three to four times annually by visiting medical geneticists, attended by patients identified by senior nurses trained in genetics. A total of 1797 patients were seen of which 94.4% were black South Africans (Christianson et al. 2000). The immense need in the province resulted in the project outreach aims being revised to the development of infrastructure. Genetically trained nursing sisters at the seven collaborating hospitals received further training in 1993 to take up this responsibility. By 1994, they were so clinically adept that common disorders were no longer referred to the visiting medical geneticists at the outreach clinics, which were reserved for cases where 'treatment was available and would significantly improve the prognosis' (Christianson et al. 2000). However, from 1994, commitment and funding to medical genetic services at the provincial level waned and eventually prevented the nurses from continuing in this function (Professor Philip Venter, Personal Communication, 20 May 2016).

Policy changes following the 1994 elections in SA resulted in genetic services being incorporated into primary healthcare countrywide. Genetic nurses were reassigned to PHC clinics where they were expected to provide both genetic and PHC services (Ehlers 2002). These changes[9] increased the workload for PHC nurses by an estimated 40% with no additional capacity (Wilkinson et al. 1997). With an emphasis on HIV/AIDS patients, all nurses were required to primarily focus on providing PHC services, to the detriment of their specialist area (Ehlers 2002). The restructuring of the healthcare system to address previous imbalances, combined with competing health needs, resulted in the depletion of posts for both nurse counsellors and medical geneticists (Beighton et al. 2012). Many genetic nurses moved into other positions or emigrated (Ehlers 2002). By 2001, only four medical geneticists, less than 20 geneticist counsellors, and an unknown number of genetic nurses remained (DOH 2001).

Although the training of community-based nursing staff was identified as a priority for the successful implementation of medical genetic services in the 2001 National Policy Guidelines for the Management and Prevention of Genetic

[8] Involving the University of the North, the University of Pretoria, National and Provincial Departments of Health, and trained nursing staff in seven rural hospitals in the Province.

[9] The provision of free healthcare to pregnant women and children under six without medical aid.

Disorders, Birth Defects and Disabilities (DOH 2001), formal training of nursing staff in medical genetics has ceased and only sporadic, ad hoc, self-funded genetics outreach continues in a few provinces.

Medical genetics in the nursing curricula

With the dismantling of the countrywide network of 16 genetic nurses in the mid-1990s, few genetic nurses and genetic nurse counsellor posts remain countrywide today.[10] With these nurses playing such a key role in genetic services, their absence is keenly felt. Research by Phaladi-Digamela to develop a competency-based curriculum framework for advanced midwives highlighted the call made by other nursing specialisations that the 'genetic nurse must come back' as they are 'better empowered in addressing genetics problems' (Phaladi-Digamela et al. 2014). This reliance on the genetic nurse stems from inadequate genetics knowledge, skills, and competencies included in basic nurse training curricula (DOH 2001). Appropriate standardised, quality content is lacking, leaving nurses ill-equipped when entering clinical practice (Secretary's Advisory Committee on Genetics Health and Society 2011). Globally, these inadequacies are preventing nurses from being prepared for their role in the new genetic era—which calls for *all* nurses to be appropriately skilled in medical genetics (Calzone et al. 2010).

Nursing education reform in SA is continuing as part of the post-apartheid transformation process with the recent incorporation of public nursing colleges into the higher education sector to comply with education legislation (DOH 2012; Armstrong and Rispel 2015; Rispel 2015). A continuing professional development (CPD) system is also being introduced for nurses, and a scope of practice is under development for the new nurse categories. However, poor governance by the main institutions involved is delaying implementation and realisation of targets outlined in the National Strategic Plan for Nurse Education, Training and Practice 2012/13–2016/17 (DOH 2012; Armstrong and Rispel 2015). This evolving nursing landscape may be an opportune time to improve the medical genetics component in nursing training.

Key genetics knowledge required by nurses should include basic scientific principles of genetics, genetic risk assessment, practice and ethics of genetic counselling, accessing genetic information resources, and when to refer, both for appropriate testing and to the medical geneticists or other specialist physicians (Lemkus et al. 1978; Alwan and Modell 1997; Penchaszadeh et al. 1999; Alwan and Modell 2003). In SA, it has been established that genetics knowledge is lacking in nursing training (Glass 2004; Prows et al. 2005; Godino and Skirton 2012; Phaladi-Digamela 2015; Rispel 2015). Genetics education in SA nursing is currently considered as 'slapdash' with a huge variation between institutions according to available facilities and staffing (DOH 2001; Phaladi-Digamela 2015). Genetics content is often superficial with little relevance to the identification of CDs, genetic counselling, or pre-natal diagnosis (Glass 2004; Prows et al. 2005; Phaladi-Digamela 2015). A study by Phaladi-Digamela in 2015 indicates that although genetics is included in the curricula of three quarters of the study participants, only 10 h or less of genetics teaching was reported by 50% of participants, falling far short of the recommended 40 h (Phaladi-Digamela 2015). The prediction by Godino and Skirton in 2012 that SA will embrace sufficient genetics in the nursing curricula by 2017 is unlikely to be achieved (Godino and Skirton 2012). Key challenges include an already full curriculum, nursing faculty/educators lacking genetic knowledge, and genetics education not being considered relevant for nurses (Glass 2004; Calzone et al. 2010; Calzone et al. 2013; Phaladi-Digamela et al. 2014).

A standardised genetic education framework for nurses is required in SA at basic and post-basic training levels incorporating both theory and clinical practice components. Such genetic knowledge is required by all nurses, including those in non-specialist healthcare, to translate genetic knowledge and technology to improve healthcare both in PHC and clinical settings (Calzone et al. 2010; Phaladi-Digamela 2015). Such an increased knowledge base could also serve as a pool from which nurses could then specialise as genetic nurses or genetic nurse counsellors.

The Medical Genetics Education Programme

Since developing such a standardised medical genetics framework is a long-term goal, an interim measure is necessary to equip nurses with genetics knowledge and skills. An existing option that could bridge this shortfall is the Medical Genetics Education Programme (MGEP). MGEP is a post-graduate distance learning, self-administered education programme originally developed in 2003 in response to a recommendation of the Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities (DOH 2001). MGEP aimed to equip registered nursing staff, particularly those involved in maternal, child, and women's health, with a comprehensive, primary healthcare medical genetic education (DOH 2001; Glass et al. 2007; Kromberg et al. 2013).

The MGEP programme was developed and piloted by a collaborative team of experts with funding from the March of Dimes (MOD) under the auspices of the Southern African

[10] Official numbers of genetic nurse and genetic nurse counsellor posts were unavailable. Three genetic nurse counsellor designated posts are known and several other nurses undertake some genetic nurse functions in non-genetic nursing posts.

Inherited Disorders Association (SAIDA), a patient advocacy support group recently relaunched as Genetic Alliance South Africa (GA-SA). It was originally intended that MGEP be implemented in two distinct parts over a period of 5 months, consisting of MGEP 1 and MGEP 2. MGEP 1 focused on theory, over a period of 4 months with one contact day per month including lectures and practical skill workshops. After the introduction of the Birth Defect Notification Tool (BDNT) in 2006 by NDOH, MGEP 1 also included basic training on completing and submitting BDNT notifications for national surveillance. Successful MGEP 1 participants could undertake MGEP 2, a 2-week course focusing on clinical diagnosis and genetic counselling (Prof Arnold Christianson, Personal Communication, June 2016). Due to funding constraints for MGEP 2, in practice, only the MGEP 1 component was taught.

Associated with MGEP is a manual *Birth Defects: Counselling and caring for children with birth defects* (Woods 2009). This distance learning tool promotes independent, home-based learning for primary healthcare professionals. The manual was developed with MOD funds collaboratively by a team of medical geneticists, reviewed by the wider medical genetics community, and edited and published by Eduhealthcare, as one in a series of self-directed learning course books [www.bettercare.co.za]. With the use of the Birth Defects Manual as a companion resource, MGEP became a successful blend of distance, self-administered home learning and face-to-face teaching.

Between 2004 and 2013, over 1000 healthcare providers (mainly labour ward nurses) were trained through the MGEP courses held countrywide, with an emphasis on rural areas (Malherbe et al. 2016). Coordinated by SAIDA with funding from the NDOH and MOD, MGEP was taught by a team of medical geneticists, genetic counsellors, and genetic nurse counsellors. It was intended that successful MGEP participants (nurses) could be further trained to assist with future MGEP teaching. This was formally piloted in Limpopo Province with genetic-trained nursing staff assisting as facilitators of a tele-teaching held MGEP course, resulting in an 86% pass rate of an MGEP 1 course (Gregersen et al. 2013).

MGEP evaluation and revision

An evaluation of MGEP was undertaken in 2007 for 96 primary healthcare nurses using a pre- and post-course questionnaire to test knowledge and skills (Glass et al. 2007). Pre-course knowledge averaged at 48% but increased to 75% post-course, and skills pre-course (e.g. drawing/interpreting a three-generation family tree) scored an average of 4.5% which rose to 86% post-course (Glass et al. 2007). The MGEP contact days of lectures and practical workshops clearly resulted in a significant improvement in skills and knowledge of participating nurses.

Widespread implementation of MGEP ceased in 2014 due to the lack of allocated government funding and of the 1000 nurses trained in MGEP, less than 100 continue to implement these skills (Malherbe et al. 2015). This has directly impacted national surveillance of CDs via the BDNT which was a key area of MGEP-trained nurses responsibility. However, some provinces, such as KwaZulu Natal, continue to implement MGEP despite the lack of dedicated funds and the absence of genetic services in the province.

MGEP is currently undergoing a process of revision under the auspices of GA-SA following a request by NDOH in 2014 for an improved medical genetics education course for healthcare professionals. The revised course will comply with Sector Education and Training Authority (SETA) requirements (six contact days over 6 months) for future SETA registration to increase the value of the course to participants and to access funding avenues. The Birth Defects Manual is also being simultaneously revised as the Congenital Disorders Course Book and will be made accessible via the open-source Bettercare website (http://bettercare.co.za) in hard copy, e-version or for free online viewing. Once finalised, both the revised MGEP course and the Congenital Disorders Course Book will be piloted and evaluated.

Future of MGEP and Birth Defects Manual

The interim use of MGEP and Congenital Disorders Course Book may be critical in developing the required genetic capacity in nursing and other healthcare professionals for the beginnings of a universal medical genetics service. To effectively implement the revised MGEP and Congenital Disorders Course Book, these tools should be integrated into the PHC streams of the National Health Insurance scheme (DOH 2015) as part of the healthcare re-engineering process. While many of the 52 District Clinical Specialist Teams (DCSTs) being established countrywide still lack specialist clinicians, the majority of nursing staff on these teams have already been appointed (Voce et al. 2014). These PHC, advanced midwifes and advanced paediatric nurses, could receive MGEP training and become genetic ‘champions’ in each district.

To successfully implement such educational programme, much may be learned from other MLIC facing very similar challenges of inadequate capacity and fragmented, heterogeneous services. The CHACO outreach project in Argentina developed a model to introduce genetic healthcare services into PHC in a province lacking genetic services by training 485 healthcare workers in genetics (Barreiro et al. 2013). The CHACO model, which was so successful that it is being implemented in four additional provinces, uses content very similar to the MGEP course and is adding a distance learning tool

(Barreiro et al. 2013). This experience highlights a number of factors to consider:

- *Pilot*, *evaluate*, *and replicate*: pilot and evaluate MGEP courses prior to scaling up, with continual monitoring and feedback to optimise content
- *Assess the local situation*: assess capacity needs in each province/district to identify participants and unique challenges in the area. In SA, an audit of genetic services being undertaken by NDOH as part of the 2001 policy revision provides an ideal starting point
- *Coordinated network approach*: building and strengthening coordination in each province/district between stakeholders and interventions. In SA, this should include the BDNT, the Perinatal Problem Identification Programme (PPIP), and the CHILD Problem Identification Programme (Child PIP) etc.
- *Sustainability*: training up local trainers to ensure continuous learning opportunities. The education and training mandate of the DCSTs ideally equips them to amplify genetic skills across other PHC streams (ward-based primary healthcare outreach teams and the school health teams) (Voce et al. 2014). Linkages with human genetics academic centres could assist in ensuring ensure quality, standardised training countrywide
- *Government and provincial commitment*: both national and provincial government buy-in are required. Scarce specialists (medical geneticists) may be introduced from elsewhere on a regular basis through outreach clinics, permanent posts created, and access to genetic technology improved. High turnover of government officials may be overcome by a provincial coordinator role
- *Hospital management buy-in*: gaining the commitment from hospital management to ensure continued implementation of the skills acquired

Future options for the MGEP course include development as an electronic tool through teaching by application on a tablet. Use of such a device would enable an array of other resources to be made available for diagnostic and treatment purposes, including a library of anonymized images to aid diagnosis, similar to the *Handbook of Genetic and Congenital Syndromes* (Winship 2003). Limited internet connectivity in rural regions could be overcome by downloading required updated resources periodically.

Conclusion

If used appropriately, widespread MGEP training could swiftly build up a nursing workforce with improved knowledge and skills in medical genetics, as has been modelled by other countries. There remains a need for a formal year-long diploma for specialised genetic nurse counsellors requiring formal accreditation by the South African Nursing Council. In the longer term, SA must follow the global examples of other regions and develop a standardised genetics education framework for integration into the nursing curricula to take advantage of the advances of genetics knowledge and technology in healthcare. With all these tools in place, the role of MGEP could then transition to that of a refresher course and ongoing, in-service training, as an option in the nursing Continuing Professional Development (CPD) system.

MGEP training could also be implemented for other healthcare professionals to bridge the medical genetics capacity deficit by ensuring doctors are also equipped with the relevant knowledge and skills to work optimally with the MGEP-trained nurses. A future goal could be to integrate MGEP content into medical school curricula, with an exit examination as a requirement for clinical qualification.

To ensure ‘no child is left behind’ in the new era of the Sustainable Development Goals (UN 2016), the potential offered by these tools must be harnessed to build up medical genetic services countrywide to improve the lives of those affected by CDs in the country.

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Compliance with ethical standards

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Conflict of interest Helen Malherbe is the Honorary Chair of Genetic Alliance South Africa (NPO 001-029). Arnold Christianson, David Woods, and Colleen Aldous declare that they have no conflict of interest.

Ethical approval This article does not contain any studies with human or animal participants performed by any of the authors.

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