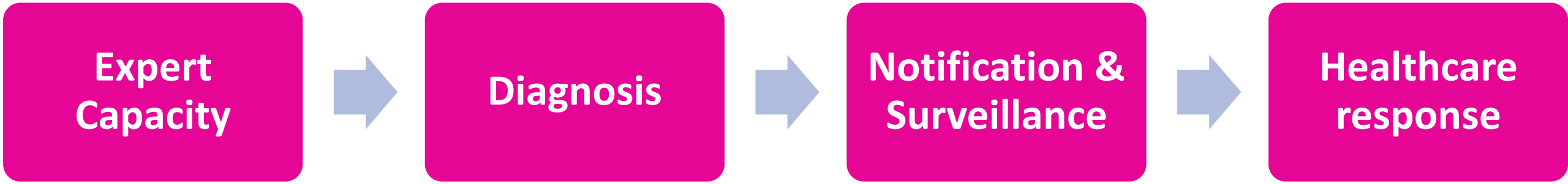
A/Prof Helen Malherbe,
Rare Diseases South Africa NPC & North-West University
27th World Congress of Epidemiology
24-27 September 2024, Cape Town, South Africa

# No surveillance without diagnosis: SA genetic services in crisis

Rare Diseases South Africa

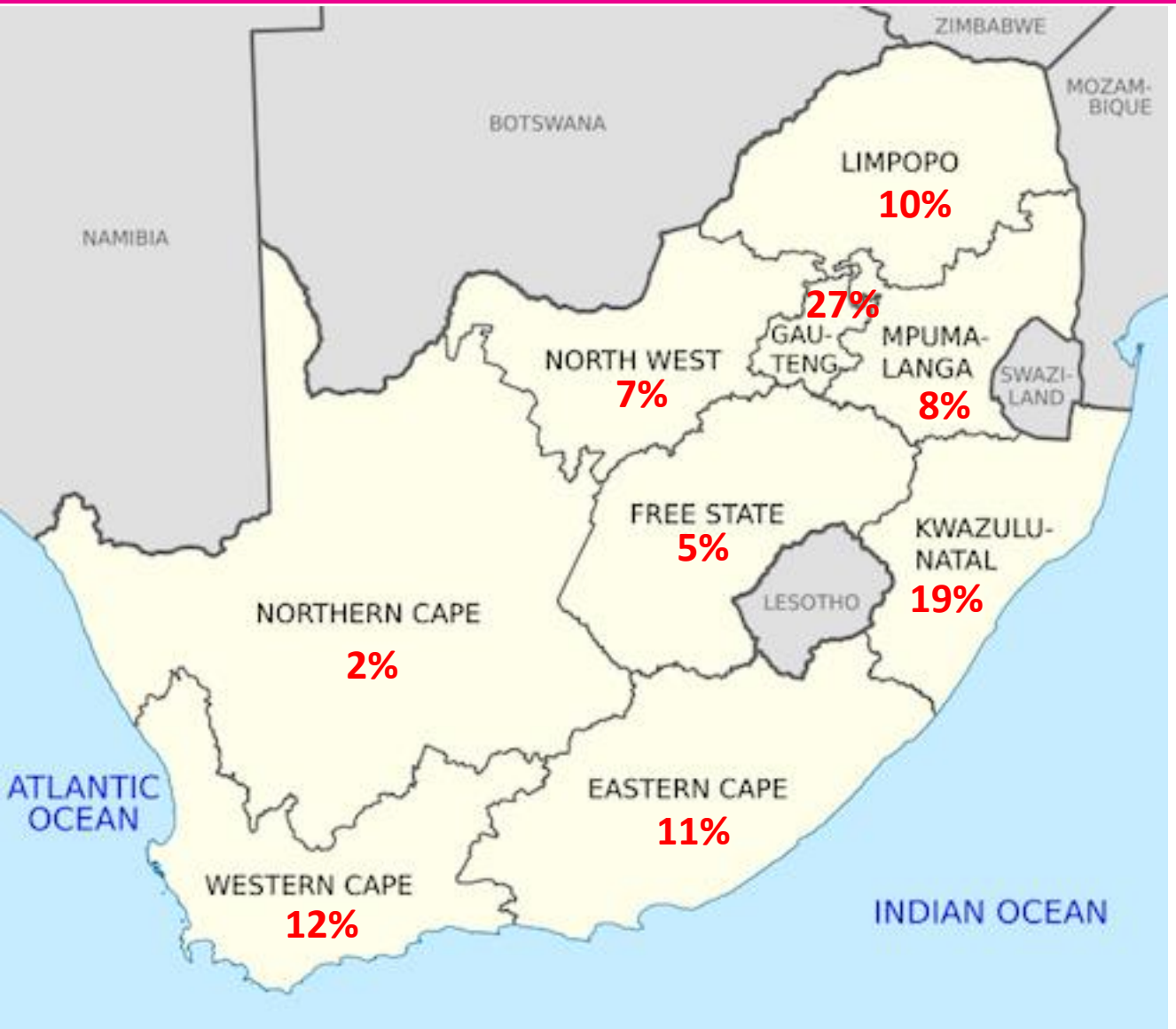
# Context: The link between surveillance and capacity

Rare Diseases South Africa




**“To count, you must be counted.**
**To be counted, you must first be diagnosed”**

# Context: South Africa




- **Population** 61m (2022) & 1.2m km$^2$
- 30% < 15 yrs & 9% >50 yrs
- **Urbanised:** 68% (2021)
- **Fertility rate:** 2.34 (2022)
- **LE** 59.3 (m) & 64.6 (f) 2021
- **Annual Births** approx. 1m P/A
- **AMA (>35 yrs):** 17% (2021)
- **YMA (<19 yrs ):** 13% (10-14: 4,042)
- **IMR:** 21/1 000 LB (2020)
- **NNMR**: 12/1 000 LB (2020)
- **U5MR:** 28/1 000 LB (2020)
- **HIV**: 22% pop or 7.7m (2022)
- **Duality**: 84% state:16% private

Adapted from: https://commons.wikimedia.org/wiki/File%3AMap_of_South_Africa_with_English_labels.svg

Stats SA & Dorrington et al (RMS) 2022

# Context: Medical Geneticists (MG)

- **Role:** Diagnose & care for those impacted by congenital disorders (CD) AKA birth defects, guide/interpret/communicate genetic tests, risk & impacts, refer etc.
- **1999/2001:** Sub-specialty - 18 medical geneticists (MG) registered with the HPCSA.
- **2007:** Designated as a 4-year primary specialty (first graduates in 2012).

**Minimum of 13 years to specialize as a MG, including:**

- 6-year medical degree (MbChB)
- 2-year internship
- 1-year community service
- 4-years of registrar training in medical genetics
- All undertaken in the public healthcare sector at 3 training centres (Wits, UCT, SU).

# Context: Genetic Counsellors (GC)

- **Role: P**rovide risk assessment and support for families diagnosed with/at risk of a CD.
- **1960's:** Informal genetic counselling services initiated.
- **1988:** Formal GC programme at Wits, Johannesburg.
- **1992**: GC formally recognised by the HPCSA (1st Wits graduates)
- **2004:** Formal GC programme at UCT, Cape Town.

**Minimum of 7 years to become a genetic counsellor, including:**

- 4-year relevant undergraduate degree with honours
- 2-year Masters in genetic counselling
- 2 -year internship (first overlaps with 2nd year of Masters)
- All undertaken in the public healthcare sector at 2 training centres (Wits, UCT).

# Aims

**The aims of this study were to:**

1. Undertake a retrospective and current audit of medical geneticists (MG) and genetic counsellors (GC) in the country to date.

2. Generate modelled prospective capacity estimates up to 2045 and compare these against recommended capacity ratios.

3. Make recommendations for addressing these quantified capacity shortages.

# Method: Retrospective & current audit (1995-2024)

Rare Diseases South Africa

## Data Gathering

- HPCSA Register
- Head of Genetic Units
- Medical Geneticists
- Genetic Counsellors
- Online resources

## Data Analysis

- Excel Workbook
- HPCSA Registration Year
- Details of Current Practice

## Results & Outputs

**PRACTISING IN SOUTH AFRICA:**

- Demographics
- Public/private
- Province

**NOT PRACTISING IN SOUTH AFRICA:**

- Emigrated
- Deceased
- Retired
- Academic/research
- Not practicing

UKZN ethics approval: BREC/00006584/202; 26 April 2024

# Method: Current Populations of MG & GC

Rare Diseases South Africa

**Training: Registrars & Interns**

**Medical Geneticists & Genetic Counsellors**

**Practicing in South Africa**

- Healthcare Sector
- Location
- Demographics

**Not Practicing in South Africa**

- Emigrated
- Retired
- Deceased
- Academic/Research
- Not-practicing

**RECORDED IN EXCEL SPREADSHEET**

# Method: Prospective Modelling (2024-2045)

Rare Diseases South Africa

## 1) Model Identification

- Wishnia J, Strugnell D, Smith A, Ranchod S. The supply of and need for medical specialists in South Africa; 2019
- 70 x HCP cadres (26 specialties/44 sub-specialties)
- Collaboration request & DTA for Python code

## 2) Model Adaptation

- 2 x HCP cadres only:
  - Medical Geneticists
  - Genetic Counsellors
- Population data & model period
- Model assumptions

## 3) Model Results & Outputs

Outputs for 4 x Scenarios:

1) Public healthcare 2025-2045
2) Private healthcare 2025-2045
3) Recommended rates
4) Adjusted Recommended Rates

Updated South African population data sourced from Thembisa version 4.7 were used.

# Model Assumptions

- ✓ **Scope: 2 x HCP cadres only (medical geneticists & genetic counsellors)**
- ✓ **Starting Population:** 2024 split by age, training period, qualifications to date etc.
- ✓ **Time period**: 2025 - 2045
- ✓ **Population Data**:
  - Thembisa version 4.7 up to 2030 (www.thembisa.org/)
  - Extrapolated 2031-2045 for public healthcare population
  - Original numbers used for private healthcare as per original model (Wishnia et al 2019)
- ✓ **Capacity Rates:**
  - Medical geneticists = 0.21 per 100 000 (NDOH 2020)
  - Genetic Counsellors = 0.84 per 100 000 (NDOH 2001, 2003)
- ✓ **Qualification Rates:** Assumes numbers remain stable as qualify ie. replaced by new registrars/interns:
  Medical Geneticist Registrars = 0.40
  Genetic Counsellors Interns = 0.33
- ✓ **Movement:**
  - Between healthcare sectors (public & private) – probabilities linked to age-ranges
  - Between Full-time & part-time - probabilities linked to age-ranges
- ✓ **Emigration:** Rate calculated using collated retrospective data (number emigrated to date divided by total number registered with HPCSA for both MG & GC).

# Modelled Scenarios

1. Used current actual rates for MG/GC for the *public* healthcare population to project rates for 2025-2045

2. Used current actual rates for MG/GC 2024 for the *private* healthcare population to project rates for 2025-2045

3. Used recommended rates from NDOH 2030 HRH Strategy for total population
   - Medical Geneticists: 0.21 per 100 000 population (NDOH 2020)
   - Genetic Counsellors: 0.84 per 100 000 population  4 x GC : 1 x MG (NDOH 2001,2003)

4. Calculated the required rate per year from 2025-2045 to reach the recommended rates in Scenario 3 by 2045 (i.e. rates of 0.21 for MG & 0.84 per 100 000 for GC)

# Results: Retrospective & Current: Medical Geneticists

Rare Diseases South Africa

41 Medical Geneticists

Registrars in-training

13 (32%) Practicing in South Africa

- 9 in PUBLIC Healthcare Sector
- 4 in PRIVATE Healthcare Sector
- Gauteng & Western Cape Provinces
- 77% female (10/13)
- 9 in training (incl. 2 supernumerary)

28 (67%) Not Practicing in South Africa

- 14 (51%) Emigrated
- 7 (25%) Retired
- 4 (15%) Deceased
- 3 (7%) Not-practicing/research etc

# Results: Retrospective & Current: Genetic Counsellors

54 Genetic Counsellors

+

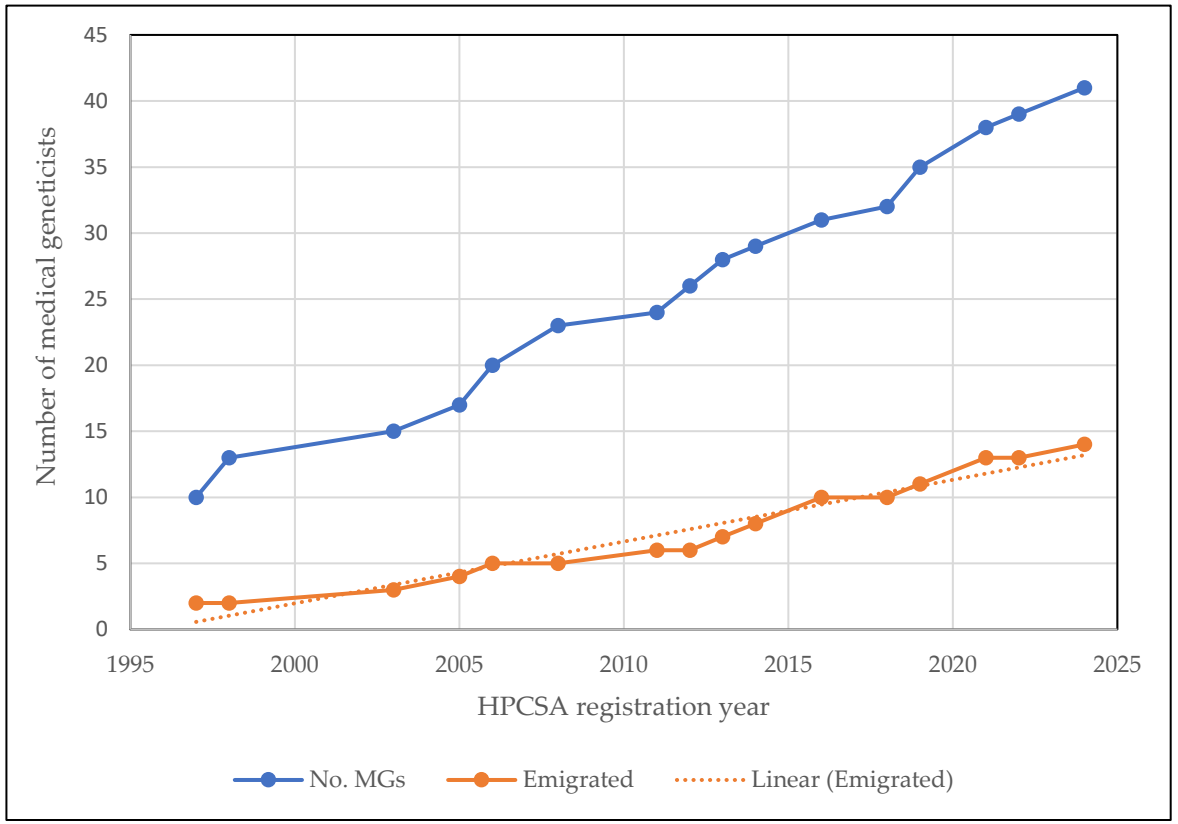
Interns in-training

28 (48%) Practicing in South Africa

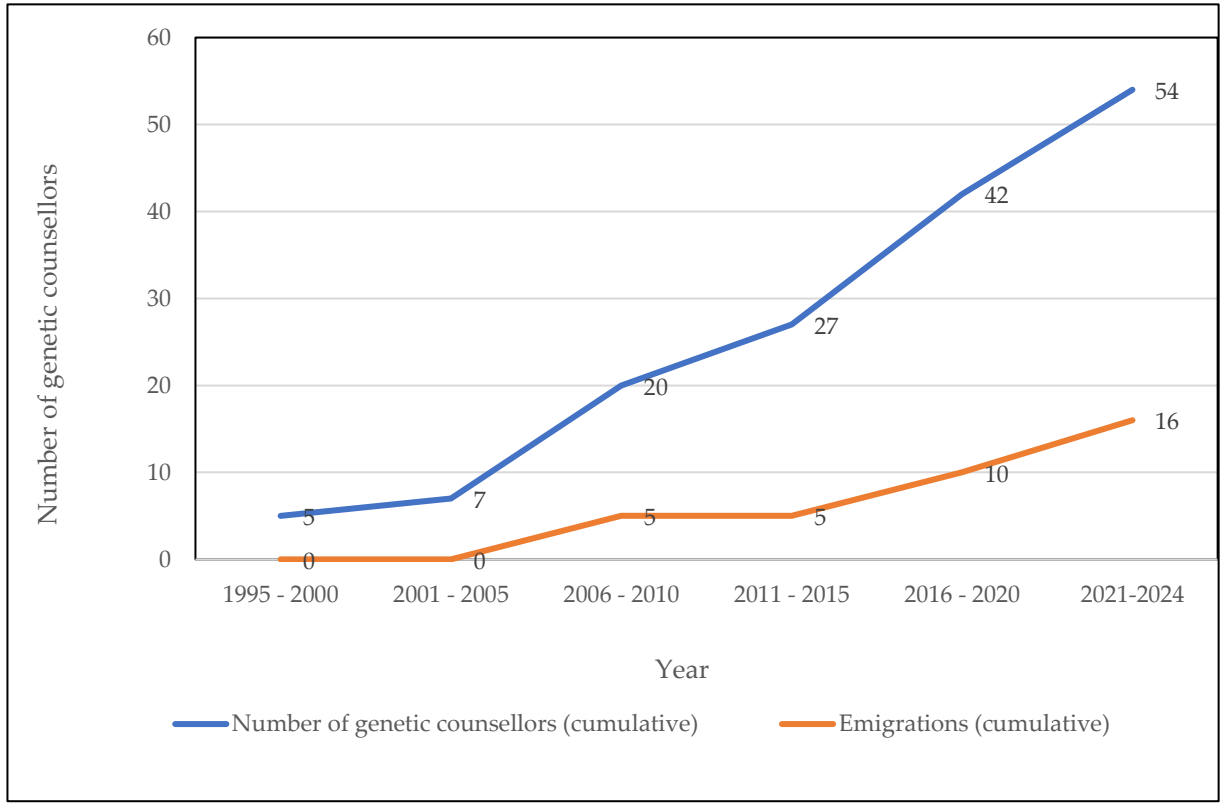
- 10 in PUBLIC Healthcare Sector
- 18 in PRIVATE Healthcare Sector
- Gauteng & Western Cape Provinces
- 96% female (27/28)
- 8 students & 8 interns

26 (67%) Not Practicing in South Africa

- 16 (62%) Emigrated
- 3 (11%) Retired
- 1 (4%) Deceased
- 5 (19%) Research/academic
- 1 (4%) Laboratory based




Cumulative number of medical geneticists who have registered in SA and emigrated (1997 to April 2024)




Cumulative number of genetic counsellors who have registered and emigrated from 1995 to April 2024

**Population & numbers of practicing medical geneticists (MG) & genetic counsellors (GC) in South Africa, 2024**

Overall:

- 8%  recc. public MG
- 2%  recc. public GC
- 20% recc. Private MG
- Publicly practicing MG & GC in WC and GP ONLY
- 22% recc. Private GC
- 6/9 provinces have no MG
- 5/9 provinces have no MG

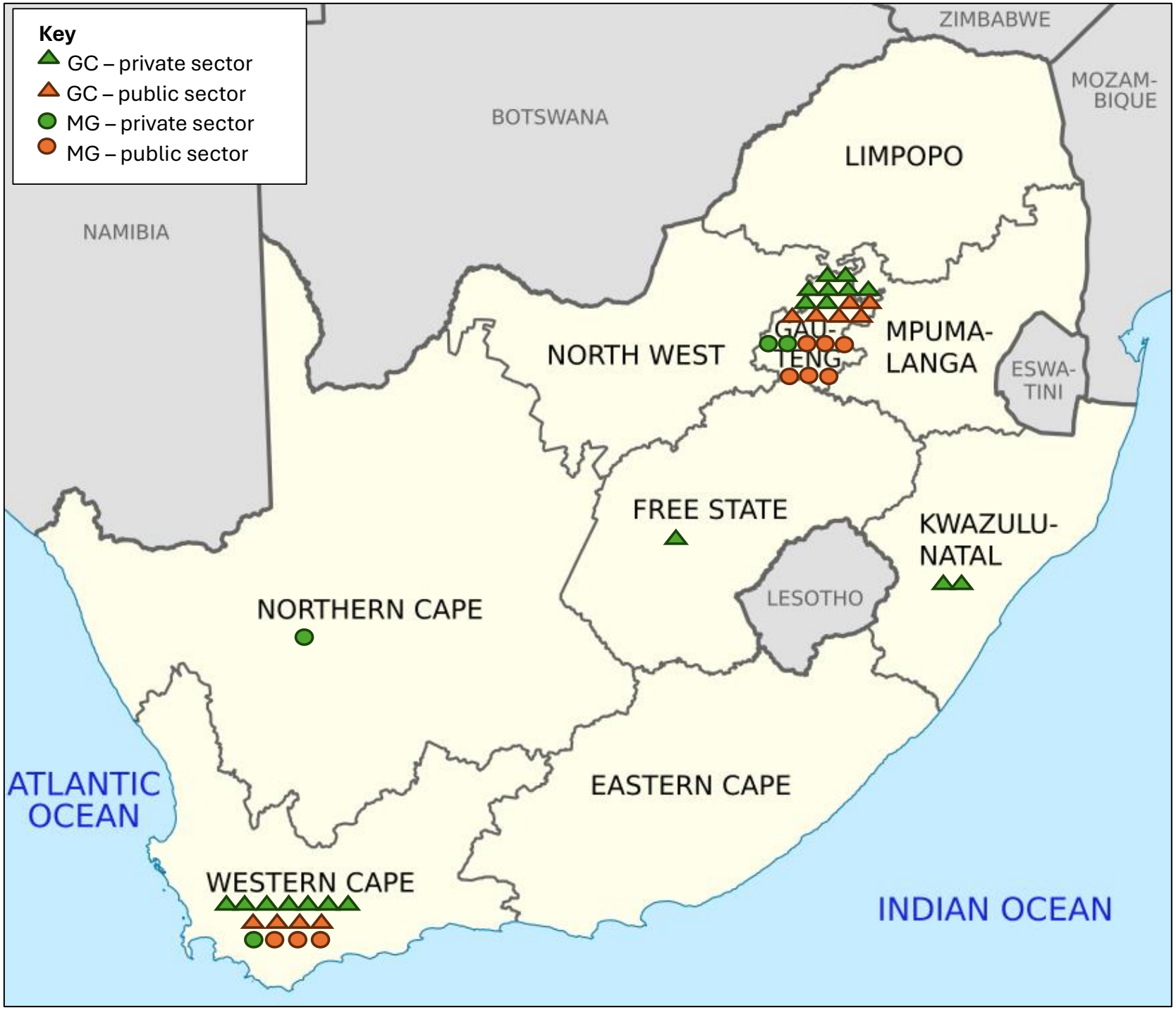
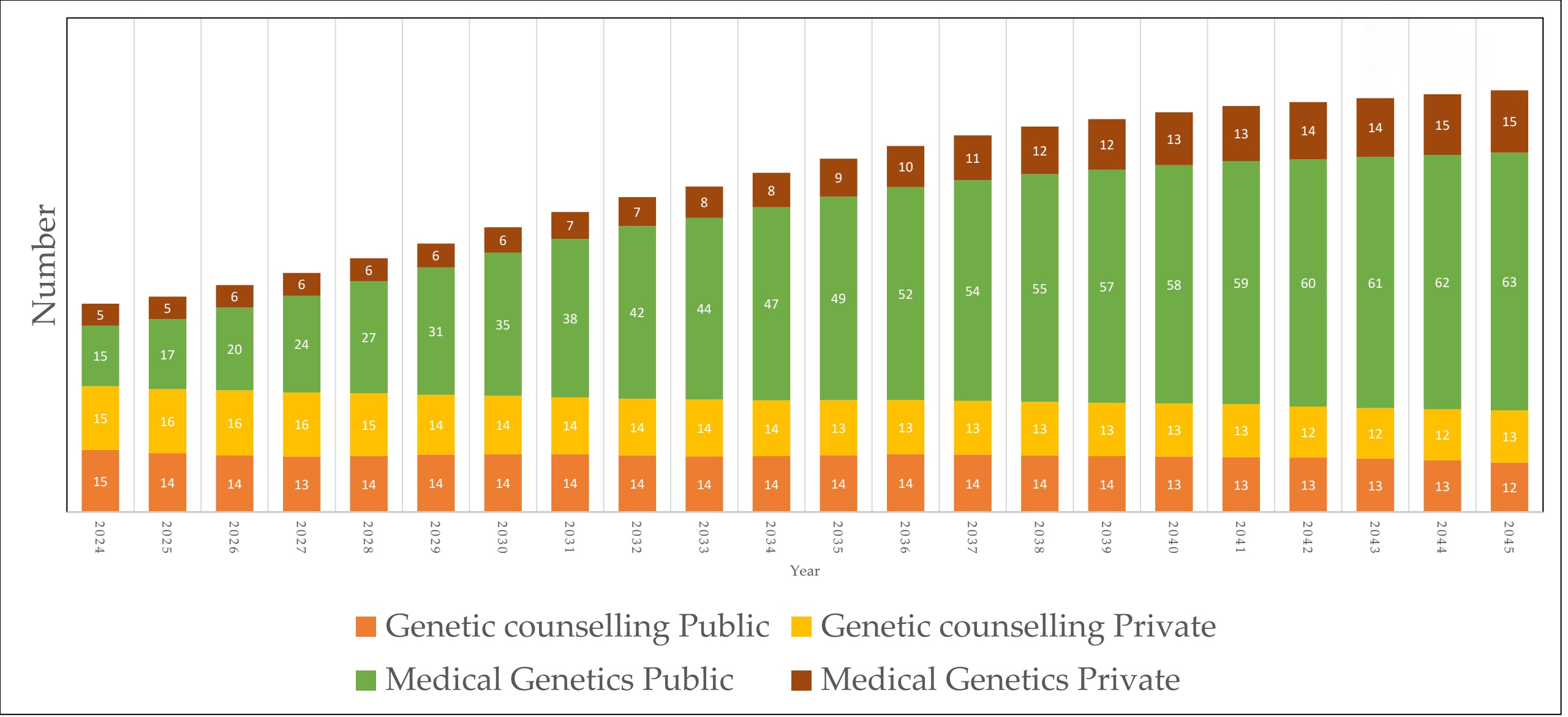
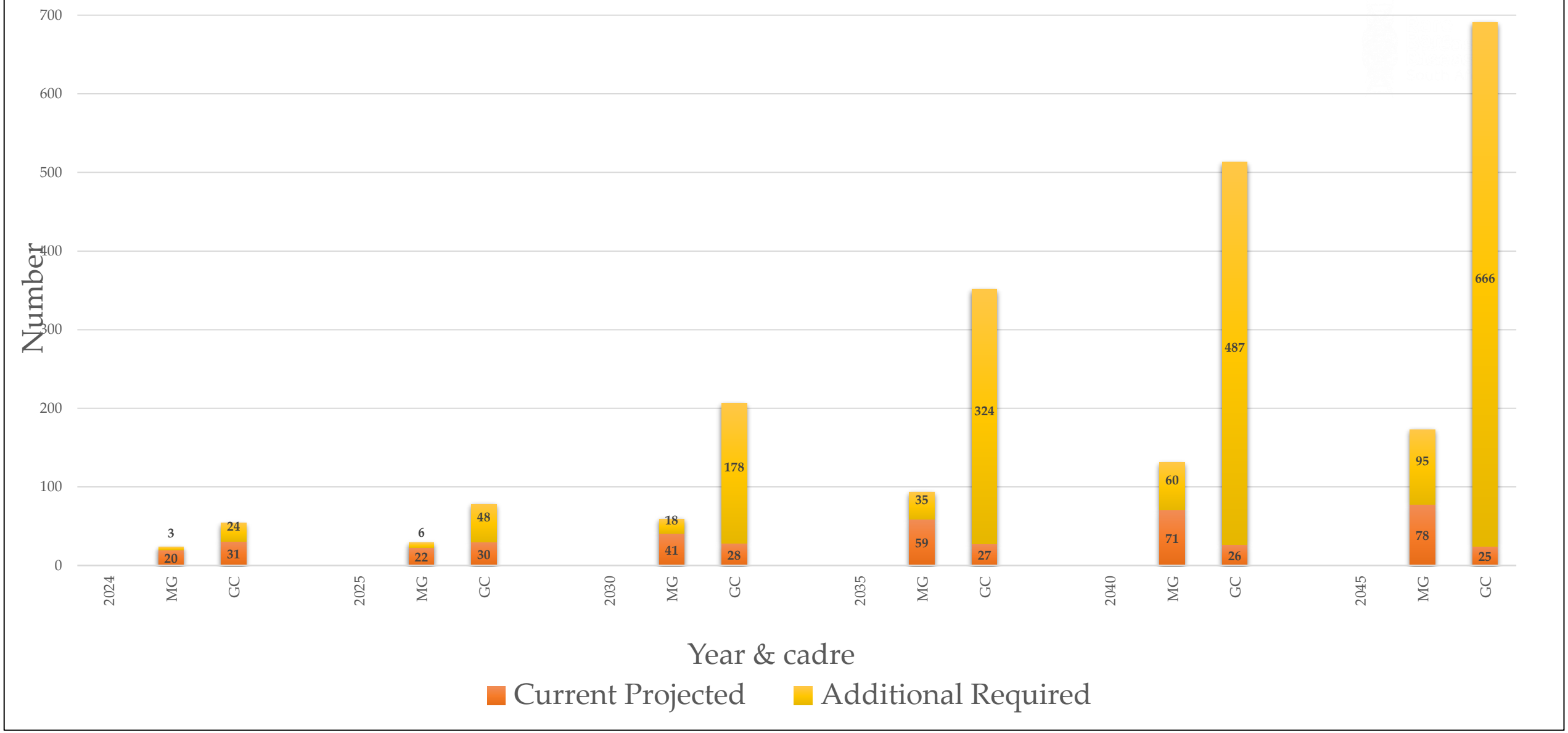



image: https://www.statista.com/statistics/1112169/total-population-of-south-africa-by-province/)




Projected growth from 2024 to 2045 in the public and private healthcare sectors for medical geneticists and genetic counsellors based on current actual (2024) rates and including medical genetics registrars and genetic counselling interns (Scenarios 1 & 2).




Current projected and additional required number of MG & GC from 2025-2045 in 5-year intervals. Data lables indicate the additional number of MG & GC required at each point in time to reach recommended capacity for each cadre by 2045.

# Discussion: Challenges to Address

**Medical Geneticists:**

- Ageing workforce
- Private practice issues
- Lack of public posts (moratorium)
- Burnout

**Genetic Counsellors:**

- Lack of appropriate/std renumeration
- Lack of public posts (moratorium)
- Emigration
- Burnout

**Common challenges:**

- Limited training capacity
- Growing health need
- Insufficient planning
- Poor implementation
- Inadequate clinical & admin support
- Under-utilization of telemedicine & outreach

# Discussion: Limitations

- **Limited to 2 x HCP cadres**: medical geneticists & genetic counsellors.
- Modelled prospective data limited by **model assumptions** - average/approximate values for projected capacity data which varies in practice.
- No comprehensive dataset of HCP in the country, necessitating **multisource** approach.
- Data used from **one year only** to calculate qualification rates – varies year on year.
- Training & employment costs for the two cadres **not included**.
- **Total** population used - stratified capacity requirements for sub-populations?

**BUT: Serves to highlight the limited capacity in genetic services in South Africa**.

# Discussion: Recommendations

**An inclusive national stakeholder commission to address capacity deficits in genetic services for effective human resource planning, including:**

- **Comprehensive** & **transparent planning**.
- **Undertake similar audits** for medical scientists, genetic nurses & other relevant cadres.
- Develop **health economics evidence-base** – what is the cost of no genetic services?
- **Clarify** & **standardise** professional level, renumeration of GC & a capacity ratio.
- **Reconsider** the role, scope and training of the genetic nurse.
- Develop **innovative** solutions, eg expand telehealth, identify funding for outreach clinics.
- Undertake **audit** of genetic/biochemical testing available in the country.

# Discussion: Conclusion

- **Insufficient** number currently practicing MG & GC (10% of required).
- **Insufficient** current/replacement training of registrars/interns to replace those lost to the system (emigration and retirement).
- **Additional** 95 practicing medical geneticists required by 2045 (plus 78 predicted).
- **Additional** 666 practicing genetic counsellors required by 2045 (plus 25 predicted).
- Current rates indicate **recommended** 2045 ratios **will not be reached** – GC declining.
- **Proposed** target capacity & progressive increases.
- Ageing population will **contribute** to the CD burden of disease.
- **Impacts** the country's ability to reach the SDG 2030 targets.

**Scale of magnitude in LMIC compared with HIC**

# References & Acknowledgements

Rare Diseases South Africa

Gomes MCM; Gomes, B.; Bailly C; McKerrow N; Christianson AL; Malherbe H.
A capacity audit of medical geneticists and genetic counsellors in South Africa, 2024 – A national crisis.
*Genes* 2024, 15, 1173. https://doi.org/10.3390/genes15091173**.**


- Dorrington, R.; Bradshaw, D.; Laubscher, R.; Nannan, N. *Rapid Mortality Surveillance Report 2019 & 2020*; South African Medical Research Council: Cape Town, 2021; p. 35.
- Johnson, L.; Meyer-Rath, G.; Dorrington, R.; Puren, A.; Seatlhodi, T.; Zuma, K.; Feizzadeh, A. The effect of HIV programs in South Africa on national HIV incidence trends, 2000-2019. *J Acquir Immune Defic Syndr* 2022, *90*, 115 – 123, National Department of Health. *Human Genetics Policy Guidelines for the Management and Prevention of Genetic Disorders, Birth Defects and Disabilities*; Department of Health: Pretoria, 2001. Available online:
- Modernisation of Tertiary Services Project Team. *Strategic Framework for the Modernisation of Tertiary Hospital Services*; 2003.
- National Department of Health. *2030 Human Resources for Health Strategy: Investing in the health workforce for Universal Health Coverage*; Pretoria, South Africa, 2020.
- Statistics South Africa. *Statistical Release P0302. Mid-year Population Estimates 2022*; StatsSA: Pretoria,
- 2022. Statistics South Africa. *Statistical Release P0305. Recorded live births 2022*; Stats SA: Pretoria, 2023.
- Statistics South Africa. *Statistical Release P0318. General Household Survey 2022*; StatsSA: Pretoria, 2023. Wishnia, J.; Strugnell, D.; Smith, A.; Ranchod, S. *The supply of and need for medical specialists in South Africa*; 2019. Available online:

# Thank You!

**research@rarediseases.co.za**

Join our community