



Feidhmeannacht na Seirbhíse Sláinte
Health Service Executive

NDTP

National Doctors Training & Planning

REVIEW OF THE
**CLINICAL
GENETICS**
MEDICAL
WORKFORCE
IN IRELAND

2019



TABLE OF CONTENTS

1	INTRODUCTION TO REPORT AND OVERVIEW OF THE CLINICAL GENETICS MEDICAL WORKFORCE	4
1.1	Introduction	4
1.2	Background to Specialty-Specific Reviews	4
1.3	Data Used and Limitations	4
1.4	About Clinical Genetics	5
1.5	The Context of Clinical Genetics in the Irish Health Service	6
2	CURRENT WORKFORCE	7
2.1	Introduction	7
2.2	Participation of Consultants/Specialists in the Medical Workforce in Ireland	7
2.2.1	The Number of HSE Approved Consultant Posts	7
2.2.2	Irish Medical Council (IMC) Data	7
2.2.3	Number of Doctors on the Specialist Register working exclusively in the Private Sector	7
2.2.4	Country of Basic Medical Qualification	8
2.2.5	Gender	8
2.2.6	Working Patterns	8
2.2.7	Permanent/Temporary Status of Consultant Contract	8
2.2.8	Age Profile of Consultants/ Specialists in Clinical Genetics	9
2.3	The Number of NCHDs Working in the Irish Healthcare System	9
2.3.1	Training Posts	9
2.3.2	Projected Training Programme Exits	9
2.3.3	NCHDs Not in Training Posts	10
2.3.4	Non-Training NCHDs Working in the Private Sector	10
2.4	Participation of non-training NCHDs in the Medical Workforce in Ireland	10
2.4.1	Country of Basic Medical Qualification	10
2.4.2	Gender	11
2.4.3	Working Patterns	11
2.4.4	Age Profile of NCHDs	11
2.5	Ratio of NCHDs to Consultants	11
2.6	Summary of Current Configuration of Clinical Genetics Workforce 2016/2017	12
3	ESTIMATION OF THE CURRENT UNDERSUPPLY OF DOCTORS IN THE CLINICAL GENETICS WORKFORCE	13
3.1	Availability of Specialist Clinical Genetics Care Services in Ireland	13
3.2	Estimation of Current Undersupply of Clinical Genetics Specialists	13
3.2.1	Contributors to undersupply	13
4	KEY DRIVERS OF CHANGE TO THE FUTURE OF THE CLINICAL GENETICS WORKFORCE	14
4.1	Development and advances in the field of Clinical Genetics	14
4.1.1	Foetal Medicine	14
4.2	Populations requirements and future burden of disease	14
4.3	Policy recommendations and proposed reconfiguration of services	15

5	CLINICAL GENETICS STAFFING IN COMPARABLE JURISDICTIONS	16
5.1	Ireland	16
5.2	Actual and Recommended International Ratio of Consultants/ Specialists in Clinical Genetics	16
6	SUMMARY	17
6.1	Participation of Consultants/Specialists and NCHDs in the Medical Workforce in Ireland	17
7	BIOCHEMICAL GENETICISTS	18
8	REFERENCES	19

1 – INTRODUCTION TO REPORT AND OVERVIEW OF THE CLINICAL GENETICS MEDICAL WORKFORCE

1.1 INTRODUCTION

National Doctors Training and Planning (NDTP) is a division within the HSE National Directorate for Human Resources, and has statutory roles in:

- Medical education and training;
- Medical workforce planning; and,
- The consultant post approval process.

Within its medical workforce planning remit, NDTP predicts and proposes on an annual basis the number of interns and medical trainees required for each specialty, as well as projecting the future medical workforce requirements for each specialty. This information then feeds into the medical education and training aspect of NDTP via the commissioning and funding of medical training required to meet workforce needs, ensuring that the training content and delivery is responsive to the changing needs of the Irish healthcare system, and supporting the retention of doctors upon completion of their training.

The main objective of NDTP is to ensure that, at all times, the Irish health service is provided with the appropriate number of specialists, who possess the required skills and competencies to deliver high quality and safe care, and whose training is matched to the model of healthcare delivery in Ireland, regardless of location.

1.2 BACKGROUND TO SPECIALTY-SPECIFIC REVIEWS

In 2014, NDTP published “Population Based Ratios of Specialists in Ireland and Internationally: An Information Source to Support Medical Workforce Planning”, which was a benchmarking exercise conducted across all medical specialties, comparing specialist numbers against international examples. Included in this benchmarking exercise were the projected numbers of specialists required per specialty over a 10-year projection timeline. Data and contextual information were requested from individual postgraduate medical training bodies and associated national clinical programmes, which were considered in the development of each specialty-specific section.

As a follow-up to this exercise, it is timely to provide a review of each medical specialty based on current available data and input again from the postgraduate training bodies and clinical programmes. These reviews are high-level and are a companion to the more in-depth specialty specific reports which are published by NDTP (i.e. “Future Demand for General Practitioners 2015-2025”, published in 2015), with workforce review reports also published for the specialties of Paediatrics and Neonatology, Palliative Medicine, Clinical Radiology, Radiation Oncology and Emergency Medicine. These reviews are useful references for those with an interest in data on the medical workforce and medical workforce planning, comprising a live repository that will be updated as each review is completed. Where a review has yet to be completed, the relevant section from the benchmarking exercise in 2014 is available for reference. All workforce planning reports are at www.hse/doctors.ie

This particular report has been informed by information submitted to the HSE National Doctors Training and Planning (NDTP) Unit by Dr Sally Ann Lynch, ICHMT, RCPI. There is no Clinical Programme for Clinical Genetics within the HSE.

1.3 DATA USED AND LIMITATIONS

The data utilised in the analysis of the medical workforce in each speciality for these reviews are drawn from multiple sources:

- HSE Doctors Integrated Management E-System (DIME), which receives data from the postgraduate medical training bodies, the Irish Medical Council (IMC) and each clinical site that employs doctors in the public health system in Ireland;
- The Irish Medical Council (IMC), Annual Retention Application Form;
- International medical workforce datasets (i.e. National Health Workforce Dataset in Australia);
- International health research groups (i.e. Health Workforce Australia).

Variations between datasets are not unexpected and therefore the results from the different sources in the reviews are **not identical**. These limitations of the datasets are due to variations in the time point of data collection, differences in the variables collected (i.e. whole-time equivalents (WTE) versus headcounts (HC)), differences in the definitions of some variables (e.g. less than full-time versus part-time), absence of variable values in datasets (i.e. missing data), and varying quality of data between sources.

The weaknesses of benchmarking domestic data against international data are known and include:

- (i) A lack of contextual consideration;
- (ii) Assumptions that the international standard is best practice; and,
- (iii) Potential complacency should the domestic value equal that of the international value.

However, there is merit in this kind of comparison as these ratios are informative in terms of contextualising the demand for consultants and specialists across international healthcare systems with similar training and healthcare delivery infrastructures to those in Ireland. Further, it provides an international baseline for comparison and can help identify areas for improvement. Irish doctors traditionally migrate to countries like the UK and Australia and so benchmarking against these countries is a useful exercise.

Should you require any further information on the reviews, please contact NDTP at doctors@hse.ie

1.4 ABOUT CLINICAL GENETICS

Clinical Geneticists deals with the diagnosis and management of inherited disorders and birth defects¹. In addition to rare genetic disorders, Clinical Geneticists also care for families with common genetic disorders and non-genetic congenital anomalies including possible teratogenic conditions.

The Clinical Genetics service at Children's Health Ireland (CHI) at Crumlin provides a diagnostic, counselling and clinical genetic testing service for children and adults affected by or at risk of a genetic condition. The service is the sole provider of general genetic counselling to the population of the Republic of Ireland. The service cares for and manages families with genetic conditions, rather than simply focusing on individual patients. Advice via telephone, email or letter without direct patient contact is also provided. The service is provided through a hub and spoke model with the hub being situated at CHI at Crumlin and spoke clinics being performed in Cork, Limerick, Galway and Children's Health Ireland (CHI) at Temple street.

Common referrals include:

- Children/ adults with congenital anomalies
- Children/ adults with intellectual disability
- Families affected by or at risk of chromosome disorders
- Families affected by inherited disorders such as Cystic Fibrosis
- Families affected by or at risk of hereditary cancer
- Families affected by or at risk of hereditary heart disease
- Families affected by or at risk of inherited neurological disorders
- Healthy family members seeking predictive genetic tests for the genetic disorder identified in their family
- Referral to advise on relevance/ significance of genetic test report
- Requests for testing in pregnancy for rare disorders (prenatal testing)
- Requests to advise on possible teratogenic malformations

1.5 THE CONTEXT OF CLINICAL GENETICS IN THE IRISH HEALTH SERVICE

The Clinical Genetics service for Ireland was founded in 1995, based in CHI at Crumlin and originally known as the National Centre for Medical Genetics. The Centre was set up to provide a service to all patients living in the Republic of Ireland. The service receives referrals from all age groups (adults, pregnant women and paediatrics) from clinicians all over the Republic of Ireland. It is predominantly out-patient based. Ward referrals for acutely ill neonates with suspected genetic conditions also form part of the remit. They work in close liaison with colleagues from molecular and cytogenetics laboratories.

The service is staffed by consultants, trainee NCHDs, non-trainee NCHDs and Genetic Counsellors. Genetic Counsellors have a science or nursing degree and have a postgraduate master's qualification in Clinical Genetic Counselling. They see patients with known genetic disorders, under the direction of a consultant Clinical Geneticist. The British Clinical Genetics society recommend 8 Genetic Counsellors per million of population². Currently the Department of Clinical Genetics has a total of 5.1 WTE Genetic Counsellors. The service is also supported by a number of administrative staff.

Cancer genetics comprises approximately 30% of the speciality's current workload. Approximately 5% of breast, ovarian and bowel cancer has a hereditary basis; in addition, there are many rare cancer susceptibility syndromes. In the era of personalised medicine, developments in Next Generation Sequencing (NGS) technology have facilitated stratification of cancers with specific treatment options following genetic tumour analysis. In addition, diagnostic and predictive testing following genetic counselling forms a core part of the work of a consultant Clinical Geneticist. The proposed model of service delivery is that of a central service hub and outreach spokes nationally³. In addition to clinic sessions in CHI at Crumlin, outreach clinics from the CHI at Crumlin department of Clinical Genetics operate in CHI at Temple street, Cork, Galway and Limerick. The main genetics specific database, known as iGene is based at CHI at Crumlin. Currently, there are approximately 35,000 family records held in the unit to date with more than 100,000 individual patients recorded on iGene.

2 – CURRENT WORKFORCE

2.1 INTRODUCTION

Clinical Genetics doctors working in Ireland work in acute hospital settings and the majority work in the public sector. Clinical Genetics is a rapidly evolving specialty and Clinical Genetics doctors are required to take account of new discoveries that may alter clinical practice¹. The population of doctors working in Clinical Genetics is made up of consultants/specialists and Non-Consultant Hospital Doctors (NCHDs). This section of the report gives a breakdown of doctors working in the specialty and NCHD posts across the public and private sectors.

2.2 PARTICIPATION OF CONSULTANTS/SPECIALISTS IN THE MEDICAL WORKFORCE IN IRELAND

2.2.1 The Number of HSE Approved Consultant Posts

Table 1 describes data from the HSE NDTP DIME (2019) database on the number of approved consultant Clinical Geneticist posts in the public health system in Ireland. Approved posts are posts that have been signed off on at the Consultant Applications Advisory Committee (CAAC). Posts may or may not be filled and as such they do not directly represent the number of consultants working in the health system at a particular point in time. As of July 2019 there were a total of 7 (7 WTE) approved posts. Three of these posts were approved in 2019 and have yet to be filled.

Table 1 HSE Number of Approved Consultant Posts as of March 2019 (Source: HSE, NDTP) ⁴		
	HC	WTE
Clinical Genetics posts	7*	7*

*A condition of one of these posts is that it may be filled by an individual with specialist qualifications in Clinical Genetics or Medical Oncology, this post is currently not filled.

2.2.2 Irish Medical Council (IMC) Data

According to the Annual Retention Application Form (ARAF) data from the IMC (2017)⁵ there were 7 doctors working in Clinical Genetics who were on the specialist division of the IMC Register. Five of these specialists were working exclusively in Ireland while two were working within and outside of Ireland. Table 2 outlines the breakdown of the Clinical Genetics specialist registered doctors participating in the workforce as per IMC ARAF data for 2017. According to these data, no Clinical Geneticists exited the Specialist Division of the register in 2017.

Table 2: Registered Doctors professionally active in the last 12 months (IMC, 2017)⁵

Table 2 Number of Doctors on the Specialist Register who Worked in Ireland in Clinical Genetics in the Previous Year (IMC, 2017) ⁵			
	Inside Ireland	Inside & Outside Ireland	Total
Number of Consultants/ Specialists in Clinical Genetics	5	2	7

2.2.3 Number of Doctors on the Specialist Register working exclusively in the Private Sector

According to the IMC's ARAF data, in 2017 there were no specialist registered doctors working exclusively in the private sector in Clinical Genetics.

2.2.4 Country of Basic Medical Qualification

Of the 7 consultants/ specialists who indicated that they were actively practicing in Clinical Genetics in Ireland in 2017, 71% qualified in a medical school in Ireland and 29% qualified in a medical school outside of the EU (Table 3). These data represent all consultants/specialists in Clinical Genetics working in Ireland to some extent and exclude those who are registered with the IMC but only work outside of Ireland.

Table 3: Country of Basic Medical Qualifications

Table 3 Country of Basic Medical Qualification of Doctors on the Specialist Register who worked in Ireland in the past 12 months in the area of Clinical Genetics (IMC, 2017) ⁵		
Qualified in Ireland	Qualified in the EU	Qualified outside the EU
5 (71%)	-	2 (29%)

2.2.5 Gender

As per the data recorded on DIME (July 2019) the headcount for consultants/ specialists in Clinical Genetics is 4. Of the consultants/ specialists in Clinical Genetics working in HSE-funded services in 2019 50% were male and 50% were female (Table 4)⁴.

Table 4: Gender Breakdown of Consultants/ Specialists

Table 4 Gender breakdown of Consultants/ Specialists in Clinical Genetics Working in Ireland 2019 (DIME, 2019) ⁴			
	Male (%)	Female (%)	Total HC
DIME, 2019	50%	50%	4

2.2.6 Working Patterns

100% of Clinical Genetics consultants/ specialists working in HSE-funded services in 2019 were working on a full-time basis (Table 5). Part-time working infers an approximate WTE rate of less than 80% (DIME, 2019).

Table 5: Working Patterns of Consultants/Specialists

Table 5 Working Patterns of Consultants/Specialists in 2019 (DIME, 2019) ⁴					
	HC	Full-time (%)	Part-time (%)	Overall WTE Rate	Total WTE
DIME 2019	4	100%	0	1.00	4

Considering the available data, it appears that the 4 consultants employed in HSE funded posts as consultants/ specialists in Clinical Genetics are working 4.00 WTE collectively⁴.

2.2.7 Permanent/ Temporary Status of Consultant Contract

All consultants/ specialists in Clinical Genetics working in HSE-funded services held a permanent contract (Table 6).

Table 6: Permanent/ Temporary Status of Consultant Contract

Table 6 Permanent/ Temporary Status of Consultant Contract 2019 ⁴			
	HC Permanent (%)	HC Non-Permanent (%)	Total HC
DIME 2019	100%	-	4

2.2.8 Age Profile of Consultants/ Specialists in Clinical Genetics

75% of consultants/ specialists in Clinical Genetics working in HSE funded services are over the age of 55⁴. Data indicate that over the next 10 years approximately 75% of consultants/ specialists are likely to exit the workforce due to retirement (table 7).

Table 7: Age Profile of Consultant/ Specialists in Clinical Genetics working in HSE funded services

Table 7 Age profile of Consultants/ Specialists in Clinical Genetics 2019 ⁴		
	50-54	55-64
DIME 2019	25%	75%

2.3 THE NUMBER OF NCHDS WORKING IN THE IRISH HEALTHCARE SYSTEM

Clinical Genetics is currently a consultant-run service. The training programme for this specialty commenced in 2013. Table 8 below outlines the number of NCHDs who retained registration with the IMC in 2017 and worked in Ireland in Clinical Genetics in the previous 12 months.

According to data from the IMC, four of the Clinical Genetics NCHDs were on the General Division of the register (Table 8) and one was on the trainee division⁵.

Table 8: NCHDs by Division of the Medical Register (IMC, 2017)

Table 8 NCHDs by Registration Type Practicing in Clinical Genetics in the Previous year (IMC 2017) ⁵				
	General Division	Supervised Division	Trainee Division	Total
NCHDs	4	-	1	5

2.3.1 Training Posts

According to NDTP DIME data⁴ there is one NCHD in Higher-Specialty Training (HST) in Clinical Genetics. The NCHD in HST in Clinical Genetics is currently in year 3 of the programme (Table 9).

Table 9: NCHDs in Higher-Speciality Training

Table 9 HSE Trainees in Clinical Genetics 2019: Distribution of posts by year of training ⁴					
	Year 1	Year 2	Year 3	Year 4	Total
HST Trainees			1		1

2.3.2 Projected Training Programme Exits

An analysis of the number of doctors in HST training currently projects that a maximum of 1 and a minimum of 0 trainees annually will complete specialist training in Clinical Genetics and be eligible for specialist registration over the next 4 years (table 14). These estimates of expected exits are pending no delays in training due to maternity/sick leave, other forms of certified leave, or year(s) out of programme.

Table 10: Projected Training Programme Exits

Table 10 Expected Training Programme Exits By Year ⁴				
	2019	2020	2021	2022
Total Trainees	0	1	0	0

2.3.3 NCHDs Not in Training Posts

As mentioned in section 2.3, of all the NCHDs who retained registration, four were on the General Division of the IMC register in 2017 (table 11).

Table 11: NCHDs Not in Training Posts (IMC,2017)

Table 11 Breakdown of Non-Training NCHDs Practicing in Clinical Genetics in the Previous Year ⁵		
	General division	Supervised division
Professionally active in past 12 months (IMC)	4	0

In contrast to the IMC data, data from HSE NDTP (DIME) shows 2 NCHDs in non-training posts as of July 2019; both of these were Registrars (Table 12). As the HSE data only captures data on doctors employed directly in publicly-funded posts, the discrepancy between IMC and HSE non-trainee number may be explained by the differences in time points of data collection and doctors working in the private sector. The IMC data records one non-training NCHD as working exclusively in the private sector (Table 13).

Table 12: Non-Training Clinical Genetics NCHDs (DIME,2019)

Table 12 Non-Training NCHDs working in Clinical Genetics ⁴			
	SHO	Registrar	Total non-training
Non-Training NCHDs	-	2	2

2.3.4 Non-Training NCHDs Working in the Private Sector

Data from the IMC (2017) infers a total of one non-training NCHD working in Clinical Genetics in Ireland practicing exclusively in the private sector (table 13).

Table 13: Non-Training NCHDs practicing in Clinical Genetics in the private sector only (IMC,2017)

Table 13 Non-Training NCHDs Practicing in Clinical Genetics in the previous year in the private sector only ⁵	
	General division
Professionally active in past 12 months	1

2.4 PARTICIPATION OF NON-TRAINING NCHDS IN THE MEDICAL WORKFORCE IN IRELAND

2.4.1 Country of Basic Medical Qualification

According to data collected by the IMC, of the total number of non-training NCHDs working in Clinical Genetics 75% qualified at undergraduate level in Ireland with 25% qualifying at undergraduate level outside of Ireland (Table 14).

Table 14: NCHDs Country of Basic Medical Qualifications (BMQ) – IMC,2017

Table 14 NCHDs Country of Basic Medical Qualification (BMQ) for NCHDs ⁵			
	Ireland	EU	Non-EU
General Division	75%	25%	-

2.4.2 Gender

According to information captured on DIME, of the Clinical Genetics NCHDs currently working in the system 50% are male and the 50% are female (table 15).

Table 15: NCHDs Gender Breakdown by Division of non-training NCHDs working in Clinical Genetics

Table 15 Gender of non-training NCHDs Working in Ireland ^{4,5}		
	Male	Female
IMC,2017	25%	75%
DIME,2019	50%	50%

The difference in the breakdown for IMC data and data captured on DIME is likely due to the difference in time points of data collection, the IMC data dates from 2017 and the DIME data from July 2019.

2.4.3 Working Patterns

Table 16: NCHDs Working Patterns Breakdown by Division of the Medical Register

Table 16 Non-training NCHDs in Clinical Genetics by Working Patterns ^{4,5}		
	Full-time (%)	Less Than Full-time (%)
IMC,2017	75%	25%
DIME,2019	100%	-

According to data from the IMC (2017)⁵ of the NCHDs on the General Division, 75% worked on a full time basis while 25% worked part time. According to data recorded on DIME, 100% of NCHDs working in publically funded posts in Ireland worked full time (Table 16).

The difference in the breakdown for IMC data and data captured on DIME is likely due to the difference in time points of data collection, the IMC data dates from 2017 and the DIME data from July 2019.

2.4.4 Age Profile of NCHDs

Of the 3 total NCHDs actively working in Clinical Genetics and captured on DIME, 33% were aged less than 35 and 67% were aged between 35-44 years (table 17).

Table 17: Age profile of non-training NCHDs working in Clinical Genetics

Table 17 Age profile of NCHDs ^{4,5}			
	<35	35-44	>55
IMC,2017	50%	25%	25%
DIME,2019	33%	67%	

2.5 RATIO OF NCHDS TO CONSULTANTS

Table 18: Ratio of all NCHDs working in Clinical Genetics to Consultants (DIME data)⁴

Total NCHD posts	N	NCHD : Consultant
Total trainees	1	1:4
Non trainees	2	2:4
Total NCHDs	3	1.25:1

The ratio of NCHDs to consultants is approximately 1.25 to 1

2.6 SUMMARY OF CURRENT CONFIGURATION OF CLINICAL GENETICS WORKFORCE 2016/2017

Table 19: Current Configuration of the Clinical Genetics Workforce

Table 19: Current Configuration of the Clinical Genetics Workforce ^{4,5}		
Stocktake of the Clinical Genetics Medical Workforce		
Assumption	Value	Source
Number of consultants working in HSE-funded services – permanent and temporary	4 HC 4 WTE	NDTP DIME (2019)
Full-time consultants in HSE-funded services	4 HC 4 WTE	NDTP DIME (2019)
Estimated number of private sector only consultants as per IMC data	0	IMC (2017)
Number of approved consultant posts for HSE-funded services	7 HC 7 WTE	NDTP DIME (2019)
Share of females in consultant employment for HSE-funded services (%)	50%	NDTP DIME (2019)
Share of males in consultant employment for HSE-funded services (%)	50%	NDTP DIME (2019)
Overall WTE rate for consultants in HSE-funded services	1.0	NDTP DIME (2019)
Exits from IMC Specialist Register	0	IMC 2017 registrations
% consultants over 55 years	75%	NDTP DIME (2019)
Total HST	1	NDTP DIME (2019)
Total non-training NCHDs	2	NDTP DIME (2019)

3 - ESTIMATION OF THE CURRENT UNDERSUPPLY OF DOCTORS IN THE CLINICAL GENETICS WORKFORCE

3.1 AVAILABILITY OF SPECIALIST CLINICAL GENETICS CARE SERVICES IN IRELAND

The core activity of Clinical Genetics service in Ireland provides:

- An outpatient based consultation service for families, run by consultant Clinical Geneticists, and genetic counsellors under consultant supervision
- Urgent inpatient consultation service and prenatal diagnostic tests for families with single gene or chromosomal disorders. Provision of clinical liaison & a gatekeeping role with the genetics laboratories and an on call consultant advisory service
- Regional outreach clinics following the European Model of Care for Genetic Services, and the Irish model of care for specialist paediatric services. Triage is performed by consultants. The numbers of patients that can safely be seen are agreed through capacity plans with clinical staff and CHI at Crumlin
- According to EU guidelines patients should receive pre-test and post-test counselling according to EU guidelines⁶, which adds to the workload of consultant Clinical Geneticists
- Under the disability act 2005, genetic testing requires informed consent about purpose and implications of the genetic test, which also impacts on the workload of consultant Clinical Geneticists

3.2 ESTIMATION OF CURRENT UNDERSUPPLY OF CLINICAL GENETICS SPECIALISTS

There is evidence of a deficit in the number of Clinical Geneticists in Ireland; this is reflected in the following:

- The service has 3,930 referrals per year. however not all referrals require attendances. Therefore, the service has 2,743 attendances a year via which it consults with approximately 5,500 patients, 58% of whom are adults. This is approximately 38% of equivalent Clinical Genetic units in the UK⁷. Considering the service is already under strain, a greater number of consultants and trainees would be required to increase the service to UK standards. Some of the reasons for these regional differences are outlined below
- Inadequate staffing levels and long waiting times are perceived as precluding referrals to the service. This indicates that Irish citizens are not getting access to genetic counselling as is described in the EU Cross border directive 2011⁸
- Currently the priority waiting list is between 15-18 months and routine referrals wait > 2 years to be seen⁹. A Clinical risk assessment associated with the waiting list is currently being performed and a number of serious adverse outcomes have been noted
- There is a lack of a co-ordinated genetic testing service in Ireland, due to funding issues in the main. A fully functioning national service cannot be accommodated. This has led to poor practice in terms of testing requests and also poor quality foreign laboratories handling Irish samples
- As the Clinical Genetics service is housed in a paediatric hospital, it has been difficult to fully support adult patients who comprise 60% of all referrals. It is also challenging for the service to support medical and surgical colleagues as the reporting lines to the HSE are through paediatric services only

3.2.1 Contributors to undersupply

- Low numbers of trainees, currently the scheme can only guarantee to advertise a training post every 4 years
- Clinical Genetics require a very good command of English and this puts off EU trainees from applying for jobs. In addition, countries such as India and Pakistan do not have training programmes in Clinical Genetics precluding recruiting from these non-EU countries
- Poor investment in Ireland is also a barrier to potential new recruits as the staffing levels and poor development of diagnostic laboratories are a deterrent
- Whilst many other specialities have had a significant increase in specialist numbers over the last decade, Clinical Genetics has not experienced the same increase. More consultant numbers in Foetal Medicine, Neurology, Nephrology, Paediatrics, Ophthalmology and Oncology have led to an increase in genetic tests being requested. However, with no increase in Clinical Genetics specialists, this has resulted in a referral bottle neck

4 – KEY DRIVERS OF CHANGE TO THE FUTURE OF THE CLINICAL GENETICS WORKFORCE

4.1 DEVELOPMENT AND ADVANCES IN THE FIELD OF CLINICAL GENETICS

- The genomics revolution, in particular the development of Next Generation Sequencing (NGS) has meant that the number of disease genes identified has increased exponentially. In tandem with the falling costs of genetic testing, this has resulted in a marked increase in the number and complexity of genetic tests being ordered by doctors from numerous specialities
- Requests for interpretation of genetic test reports are an increasingly common reason for referral reason as many more tests are being ordered. Due to service constraints there are clinicians who don't seek Clinical Genetics services and are dealing with complex reports without specialist support. This is a cause of concern.
- The role of the Clinical Geneticist is evolving internationally with the development of Multi-Disciplinary Teams (MDTs) where individual consultant Clinical Geneticists have specialist expertise in a key specialist area (e.g. Cardiac, Cancer, Neurology, Ophthalmology) and support that speciality with variant interpretation at MDT meetings. The MDT approach is a major driver in the development of the Clinical Genetics specialty¹⁰

4.1.1 Foetal Medicine

- Developments in the field of Foetal Medicine require Clinical Genetics support. The development of Non Invasive Prenatal Testing/ Diagnosis (NIPT/NIPD) techniques has fundamentally altered services offered to patients in and the options available to them in future pregnancies.
- In addition, antenatal Comparative Genomic Hybridisation (CGH) array is being offered without any Clinical Genetics support because of a lack of capacity. This has been highlighted as an area of concern but the service lacks capacity to take on this additional workload
- Antenatal exome testing is available internationally and will require planning in Ireland. The shortage of consultant Clinical Geneticists has meant these investigations are being introduced without support from the current service which is a concern. In a highly sensitive area, it is critical that parents get impartial expert advice on complex genetic test reports. This is particularly important as some couples will opt to terminate the pregnancy and it is critical that prior to a couple making this decision, that they have had access to accurate information regarding the likely significance of the genetic test result on the health of the foetus. Currently parents are being directed to services in the UK

4.2 POPULATIONS REQUIREMENTS AND FUTURE BURDEN OF DISEASE

- Ireland has one of the highest birth rates in the EU at 13.1 per 1,000 (2018)¹¹
- Ireland has an endogamous population; Irish Travellers who have >100 rare genetic disorders which have recently been categorised¹². They have a high incidence of genetic disease and attend Clinical Genetics clinics on a disproportionately greater rate than the general population¹³
- Cascade screening has increased and now accounts for approximately 12% of all Clinical Genetics referrals. Cascade screening is where at risk relatives of an affected individual request genetic testing to determine their risk. A recent audit of cascade screening in the inherited cardiac condition showed that for every 1 individual who tests positive, 6 relatives request testing⁷. Irish families being large mean that cascade screening involves larger numbers of relatives in comparison to the UK⁷
- Approximately 6% of any population will suffer from a Rare Disease (RD) in their lifetime¹⁴ and as 80% are genetic in origin these are responsible for approximately 80% of referrals. This means that approximately 225,600 Irish patients are currently living with a genetic RD¹⁴

4.3 POLICY RECOMMENDATIONS AND PROPOSED RECONFIGURATION OF SERVICES

- The National Plan for Rare Diseases which was launched in July 2014, is highly relevant to Clinical Genetics,¹⁴ it makes the following recommendations:
 - o With respect to pregnancy, where family members are known to be at risk of being carriers of genes for rare diseases, that they have appropriate access to pre-conception genetic testing and counselling, which can inform them about the risks involved in becoming pregnant
 - o Governance arrangements for 'send out' genetic tests need to be strengthened
 - o The National Plan for Rare Diseases outlines the need for National Centres of Expertise (CoEs) in Ireland to be identified for groupings of rare conditions, based on clinical need and built on foundations already established. There is an urgent requirement for the HSE to map out CoEs and healthcare pathways, and to acknowledge the different role and competencies of CoEs and centres providing care at local level, such mapping to be aligned with the re-organisation of Irish hospitals into hospital groupings
- Both the European recommendation on Rare Diseases¹⁵ and the 2011 EU Directive on patients' rights in cross-border health-care [Article 12 & 13]⁸ encourages measures to raise awareness among health professionals of diagnostic tools for rare diseases, and clarify the possibility for referral of patients to other EU countries
- According to the EU Directive on patients' rights⁸ member states are being encouraged to identify centres of expertise for rare disease. The Clinical Genetics Centre has only been partially recognised, as they do not have the multidisciplinary team in place to fulfil the criteria laid down by this directive. This means they have not been able to become members of relevant European reference networks, which may impact their ability to get timely access to diagnosis and treatments for patients

5 – CLINICAL GENETICS STAFFING IN COMPARABLE JURISDICTIONS

5.1 IRELAND

Ireland's actual ratio of consultant Clinical Geneticists per 100,000 has been calculated using NDTP DIME data which indicates a consultant headcount of 4. This equates to 0.1:100,000 of the population (see Table 21). The Royal College of Physicians, UK recommend a ratio a ratio of 0.3:100,000 which equates to 15 HC. The Royal College of Physicians of Ireland (RCPI) endorses this recommendation.

Table 20: Actual & Recommended Ratio of Consultant Clinical Geneticists per population – Ireland¹⁶

Table 20 Ireland Actual & Recommended Ratio per population ¹⁶		
	Actual	Recommended (2017) ¹⁶
Ratios – Ireland	4	15

5.2 ACTUAL AND RECOMMENDED INTERNATIONAL RATIO OF CONSULTANTS/ SPECIALISTS IN CLINICAL GENETICS

NDTP acknowledges that there are shortcomings related to the use of direct comparisons and benchmarking of international ratios of doctors to the population (across countries and service deliveries, for example) in order to determine the appropriate numbers for Ireland. NDTP endorses an in-depth examination of service delivery and related workforce requirements based on policy, new models of care development, population change, epidemiological trends, technological and economic trends among other things in the development of workforce plans for medicine.

However, direct population-based comparisons are useful in reviewing how Ireland compares with international jurisdictions where service delivery and medical training models are comparable. Table 21 compares the current and recommended ratios of Clinical Geneticists per 100,000 of the population. Ireland has a lower ratio of Clinical Geneticists per head of population than England, Scotland and Australia and is below the recommended ratio of 0.3:100,000 of population¹⁶.

Table 21: Comparison of total ratio per 100,000 of the population

Table 21 Total Ratio per 100,000 of the Population				
Consultants/Specialists Clinical Genetics	Ireland	NHS England	NHS Scotland	Australia
Current	0.1 ⁴	0.2 ¹⁷	0.37 ¹⁸	0.2 ¹⁹
Recommended	0.3 ¹⁷	0.3 ¹⁷	0.3 ¹⁷	0.3 ²⁰

6 – SUMMARY

6.1 PARTICIPATION OF CONSULTANTS/SPECIALISTS AND NCHDS IN THE MEDICAL WORKFORCE IN IRELAND

- Permanent/Temporary Status of Consultant Contract: In HSE-funded services 100% held a permanent contract
- Working Patterns: For specialists, 100% were working on a full-time basis, and 100% of NCHDs worked full-time
- Gender: For specialists, 50% were male and 50% were female, while for NCHDs 33% were male and 67% were female
- Age: For specialists, the majority were between the ages of 55-64 years old (75%), while 25% were between the ages of 50-54 years. For NCHDs, 33% were aged between 30-35 years 67% were aged between 35-44 years
- Country of Basic Medical Qualification: For specialists, 71% qualified in a medical school in Ireland and 29% qualified outside of the EU. For NCHDs 75% qualified in a medical school in Ireland and 25% qualified in a medical school in the EU but outside of Ireland
- Private practice: No specialist registered doctor in Clinical Genetics (of a total of 7) was working exclusively in the private sector. One non-training NCHD recorded working exclusively in Clinical Genetics in the private sector on a part time basis (<10 hours per week)
- Expected Training Programme Exits: One current trainee will complete their HST Clinical Genetics training and be eligible for specialist registration over the next 5 years
- Submissions made to NDTP by Dr Sally Ann Lynch, on behalf of the ICHMT, RCPI, infers a significant shortage in the number of Clinical Geneticists working the Irish health care system i.e. 4 consultants working in the system today versus an actual demand for 15 consultants

7 – BIOCHEMICAL GENETICISTS

There is an element of overlap between the Clinical Genetics specialty and Biochemical Genetics which is the branch of medicine concerned with the study of inborn errors of metabolism, the diagnosis, genetic counselling and management of individuals of all ages with inherited metabolic diseases (IMDs).

The National Centre for Inherited Metabolic Diseases (NCIMD) is based at CHI at Temple Street and the Mater Misericordiae University Hospital, attending to children and adults, respectively. The children's services are staffed by Paediatricians with a Special Interest in Metabolic diseases, whilst the Adult Services are under the supervision of Clinical Geneticists with Specialist training in Biochemical Genetics.

However Biochemical Geneticists are not employed as such, they are contracted as Paediatricians or Metabolic Disease specialists, and these are covered under other specialty workforce planning and are not included as part of this report to avoid reporting on those employed in this area twice.

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