

*D.E. Barton*

## Genetic Services in Ireland

National Centre for Medical Genetics,  
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### Country Background: Demography, Geography and Infrastructure

[Source for this section: Irish Government Web site (www.irlgov.ie)].

The Republic of Ireland is comprised of 26 of the 32 counties which make up the island of Ireland. Many different groups of immigrants (Celts, Vikings, Normans, English) have contributed to its present population. In 1841, the area comprising the present Irish State had a population of over 6.5 million. A combination of famine and emigration reduced this to 2.8 million by 1961, but since then immigration and natural growth have brought the population back up to 3.7 million (1995 census). The major centres of population are Dublin (915,000), Cork (174,000), Limerick (75,000) and Galway (51,000). 59% of the population live in cities and towns of 1,000 people or more. The overall population density is 51 persons/km<sup>2</sup> with large variations between the east and south, where densities are highest, and the less populous west of the country.

A high proportion of the population is concentrated in the younger age groups. Approximately 43% of the population is under 25 and approximately 27% is under 15. In 1993 for the first time on record, the birth rate fell below the minimum population replacement rate of 2.1 births per woman during child-rearing age, to 1.93 births per woman. Total births in 1994 were 47,929 compared to a peak of 74,064 births recorded in 1980. However, 1996 figures indicate a recovery to 50,358 births – still below the replacement rate, but indicating that the decline has been reversed.

Freedom of conscience and the free profession and practice of religion are, subject to public order and morality, constitutionally guaranteed. The majority of the people belong to Christian denominations. At the 1991 census, approximately 92% of the population of the Republic of Ireland were classified as Roman Catholic.

### Services – Health and Social Security

The health system is primarily funded from general taxation and is publicly provided, although private health care retains a considerable role. Public health expenditure in 1994 amounted to IR £ 2,085 million or 5.9% of GDP. Private health expenditure was estimated at a further IR £ 660 million or approximately 2% of GDP. This latter section is primarily funded through the non-profit-making Voluntary Health Insurance scheme, which covers approximately one-third of the population. The scheme is based on community rating (no weighting of premiums for age or health status). The British health care company BUPA has recently (January 1997) entered the Irish market on the same basis. Everybody ordinarily resident in Ireland has either full or limited eligibility for the public health services; the type of eligibility is determined by a means test. Approximately one-third of the population has full eligibility and these have free access to the complete range of services. The remainder of the population have limited eligibility; this entitles them to consultant and hospital care subject to small charges. They obtain GP services and prescribed drugs privately. They can avail themselves of refund schemes in cases of excessive expenditure on prescribed drugs.

**Table 1.** Personnel in medical genetics in Ireland

| 1996 data             | National centre | Elsewhere (estimate) | Total   |
|-----------------------|-----------------|----------------------|---------|
| Clinical geneticists  | 1               | 1                    | 2       |
| Genetic nurses        | 1               | 0                    | 1       |
| Cytogeneticists       | 6.6             | 7                    | 13.6    |
| Molecular geneticists | 4               | about 4              | about 8 |

**Table 2.** Laboratory genetic services in Ireland

|                           | Molecular genetics | Cytogenetics | Biochemical genetics |
|---------------------------|--------------------|--------------|----------------------|
| University College Galway | yes                | yes          | no                   |
| National Screening Centre | yes                | no           | yes                  |
| University College Cork   | yes                | no           | no                   |
| Trinity College Dublin    | yes                | yes          | no                   |

The health strategy 'Shaping a Healthier Future', published in 1994, aims to enhance the health and quality of life of the population. In future, it is intended to assess the health care system by its effectiveness, i.e. the benefits received by the users of services, rather than by levels of service. A high degree of emphasis is placed on preventative medicine.

### Dimension 1: Availability

State-funded genetics services are delivered through the National Centre for Medical Genetics. The budget for genetics services in 1995 was IR £ 550,000. The National Centre for Medical Genetics, founded in 1994 on the basis of a Department of Health report, is the body charged with delivery of state-funded medical genetics services. A comprehensive service is offered by laboratories (see below).

Table 1 summarises the personnel available for the delivery of genetic services in Ireland and the range of tests available in several laboratories offering specific genetic tests on a fee-per-sample basis are shown in table 2. There is one clinical geneticist in private practice. Newborn screening for PKU, hypothyroidism and galactosaemia is provided by the National Newborn Screening Centre at The Children's Hospital, Temple Street, Dublin.

### *The National Centre for Medical Genetics*

*Structure.* The National Centre for Medical Genetics provides a comprehensive service for all patients and families affected by or at risk of a genetic disorder. Although the Centre is based in Our Lady's Hospital for Sick Children, Crumlin, it is a national centre for both children and adults. The Centre comprises three units: a clinical service, a cytogenetics laboratory, and a molecular genetics laboratory. There is a strong research element to the unit. The University College Dublin Department of Medical Genetics is based in the National Centre for Medical Genetics. The National Centre for Medical Genetics liaises closely with the National Newborn Screening Unit at the Temple Street Hospital. Because the Centre is fully funded by the Department of Health (through Our Lady's Hospital for Sick Children), no charges are made for any of the services offered.

*Clinical Genetics Service.* The clinical genetics service has approved posts for a consultant clinical geneticist, a registrar, a nurse counsellor and a secretary. Due to the departure of the original consultant, only the secretarial post is filled at present, but all of the other posts are expected to be filled in 1997. It is also expected that a further consultant post and support team will be approved in 1997.

*Cytogenetics Service.* The cytogenetics laboratory provides a comprehensive chromosome analysis service covering blood, bone marrow and skin, and placental fibro-

blast and amniotic fluid cultures. Newer techniques such as FISH and CGH, for diagnosis of microdeletion syndromes and for the detection of aneuploidy from interphase cells, are routinely provided where appropriate. The major work of the laboratory is the analysis of samples from individuals with suspected constitutional chromosome anomalies. A cytogenetic service is also provided for acquired diseases, in particular leukaemias and lymphomas, where a cytogenetic abnormality can have clear prognostic significance.

*Molecular Genetics Service.* The molecular genetics laboratory is the only laboratory in Ireland which provides a molecular diagnostic service for a wide range of inherited disorders. The laboratory continues to introduce new tests as they become available. The major work of the laboratory is the diagnosis of single-gene disorders. Many adult-onset conditions are covered by the laboratory, including many neurological disorders and inherited forms of cancer. A bank of DNA representing every sample received by the laboratory is maintained and cross-indexed to the patient register.

*Department of Medical Genetics, University College Dublin.* The University College Dublin Department of Medical Genetics is based in the National Centre for Medical Genetics. Much of the research programme of the department is based on the clinical cases with which the Centre has been involved. This close association allows for the rapid transfer of new research findings into practical genetic testing of benefit to patients. The National Centre for Medical Genetics can provide the opportunity for other Irish research laboratories to transfer their research findings into the clinical setting. The Centre participates in teaching a course for undergraduate medical students at University College Dublin and at the Royal College of Surgeons of Ireland. In addition, members of the unit provide teaching to postgraduates of many disciplines, as well as lay groups and patient support groups.

#### *Recognition/Accreditation*

Medical genetics in Ireland is still in its infancy, and there is no specific recognition of genetics as a clinical specialty. Geneticists in Ireland look to the UK (and the USA) for training, accreditation and quality assurance schemes.

## **Dimension 2: Access**

Genetic services provided by the National Centre for Medical Genetics are free of charge to the patient. All other genetic services described above and in the tables are charged for on a fee-per-sample or fee-per-session basis. As genetics has not in the past formed a major part of the curriculum in Irish medical schools, knowledge of genetic services and how they may be used is very patchy. This situation has been exacerbated by the absence of any organised medical genetic service. Both of these areas have seen major advances with the opening of the National Centre for Medical Genetics and the UCD Department of Medical Genetics, and it is expected that knowledge of genetics amongst general practitioners and specialists will improve rapidly.

#### *Religious and Legal*

[Source for this section: Irish Government Web site ([www.irlgov.ie](http://www.irlgov.ie))].

The vast majority of Irish citizens describe themselves as being Roman Catholics, and this church has an important influence on issues of morality. Increasingly, however, a more tolerant 'pluralist' ethos is emerging, especially in the large urban centres. Legislation concerning abortion has changed radically in recent years, with Supreme Court rulings that abortion is permissible in cases where there is a clear danger to the life of the mother, but not otherwise. Following an 'Abortion advice and information' referendum in 1994, legislation was introduced to allow doctors to give patients information about abortion. This legislation was referred to the Supreme Court, which found that while a doctor must not advocate or promote the termination of her pregnancy, he/she may give a woman information on abortion services lawfully available outside the State provided that it is given in the context of full information, advice and counselling on all courses of action open to her, including abortion. The final decision, on the course of action to be taken, is left to the woman. A doctor cannot make an appointment with an abortion service on behalf of a woman but once it is made he/she may communicate in the normal way with another doctor with regard to the condition of his/her patient, provided that such communication does not in any way advocate or promote the termination of pregnancy. This is now the law.

A debate is going on within the Irish medical community as to what obligations a clinician has to inform a patient about services to which they have a moral objection. While all agree that clinicians cannot be forced to participate in any procedure or treatment to which they

are opposed on moral grounds, some go further and say that clinicians are not bound to make patients aware that such procedures or treatments exist, while others maintain that clinicians should be obliged to make a referral to a clinician who can provide the treatment in question. An Irish medical conference passed a motion in 1996 condemning prenatal diagnosis.

Amniocentesis is carried out at several maternity hospitals in Dublin and elsewhere, but patients may have difficulty in obtaining a referral to one of these hospitals, depending upon the position taken by their primary care clinician.

### **Dimension 3: Life Sustaining**

[Source for this section: Irish Government Web site (www.irlgov.ie)].

Life expectancy is 72.2 years for men and 77.7 years for women. Premature deaths arise mainly from what are often referred to today as 'diseases of civilisation'. These include cardiovascular disease and cancer. At 361 deaths per 100,000 of population in 1991 the rate of death from cardiovascular disease is higher than the average in the EU. The rate of death from stroke has fallen to the EU average in recent years but the rate of death from heart attacks, while falling also, is still above average. The death rate from cancer is 245 per 100,000 and is increasing. This is above the EU average.

Perinatal and infant mortality in Ireland are now at the lowest levels ever achieved (Ireland compares very favourably with other developed countries), and the decline can be expected to continue, though perhaps at a less dramatic rate. For the most recent available year (1991), the perinatal mortality rate stood at 9.4 deaths per 1,000 live and still births. Ireland has an infant mortality rate of 5.9, although the infant mortality rate for the Travelling Community is significantly above the national average. These reductions in mortality have been achieved through improvements in living conditions as well as in the standard and uptake of maternal care. Birth rates in Ireland have been falling quite rapidly since 1980. Irish birth and fertility rates are now approaching those of other EU countries.

The National Centre for Medical Genetics is actively building contacts with cancer specialists in other hospitals with a view to co-ordinating patient care. However, no formal network is in place at present.

### **Dimension 4: State of the Art**

There are no published recommendations for teaching or training in genetics, or for minimum laboratory equipment. Again, Ireland would look to the UK and the USA for such guidelines, and attempt to distil the best from each.

Basic genetics is taught at undergraduate level to science and medical students in all Irish universities. The medical genetics content of such courses varies widely. Trinity College, Dublin University runs postgraduate courses on 'genetics and molecular biology in clinical medicine', and has recently introduced an MSc course in this area for clinicians. The author is not aware of any other relevant courses.

### **Dimension 5: Non-Harmful**

The National Centre for Medical Genetics prepares an annual report on its activities for the Department of Health.

The Molecular Genetics Laboratory at the National Centre for Medical Genetics participates in the UK NEQAS for molecular genetics. The Cytogenetics Laboratory at the National Centre for Medical Genetics participates in the UK NEQAS for Cytogenetics and the QA scheme run by the American College of Pathologists. Other genetics laboratories in Ireland do not participate in quality assurance schemes, as far as the author is aware. Again, Ireland would look to the UK and/or the USA for accreditation of laboratories and approval of training centres.

### **Dimension 6: Effectiveness**

There are currently no data published on outcomes, and no desired outcomes have been formulated or agreed.

### **Dimension 7: Consumer Satisfaction**

There are public debates on issues related to genetics, such as the availability of abortion, amniocentesis, in vitro fertilisation and labelling of genetically engineered food as well as the debates within the medical community described above. There are no published reports on consumer satisfaction with the genetics service.

Influential patient groups include the Huntington's Disease Association and Muscular Dystrophy Ireland. Such groups are represented by an umbrella group known as the Inherited Diseases Organisation, which lobbies the Government on issues of interest to those with inherited disorders, including the provision of medical genetics services.

#### **Publications Related to Genetic Services**

- 1 Report of the Committee to Examine Medical Genetics Services (1990) by the Department of Health.
- 2 The National Centre for Medical Genetics reports to the Department of Health annually via its host institution.

#### **Irish Society for Human Genetics**

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#### **Validator**

The National Centre for Medical Genetics is the only official body representing medical genetics in Ireland, therefore the chapter has not been sent elsewhere for validation.