

LEADING ARTICLES

Genetics Surveillance in Victoria: A Recent Review

Christine A Stone and Rosemary Lester

ABSTRACT

New techniques in genetics surveillance have facilitated an improved public health approach to the detection of, and interventions offered for, a range of important genetic conditions. Over the past ten years, scientific advances associated with genetics have been increasing at an explosive rate. This has meant that an increasing number of diagnostic, predictive and carrier tests are available. This article describes the prevalence of some genetic disorders, the testing activity associated with those conditions and the links with public health.

INTRODUCTION

Surveillance for birth defects and genetic diseases started in the late 1950s and early 1960s as a response to the links between thalidomide (an anti-nausea drug) and severe birth defects.¹ Newborn screening for phenylketonuria at a population level started in the late 1960s and was soon followed by prenatal genetic diagnosis for chromosomal and biochemical disorders. The identification in 1989 of the cystic fibrosis (CF) gene and its most common mutation immediately raised the possibility of CF carrier detection by DNA analysis. In the 1990s, testing for single gene disorders commenced.

New advances in genetics promise many new diagnostic, prognostic and possibly curative technologies. These have far reaching implications. As Director of the Office of Genetics and Disease Prevention at Centers for Disease Control and Prevention, Atlanta, Georgia, Muin Khoury states "There will be unavoidable integration of new genetic information in all public health programs and across all diseases, whether or not the diseases are labelled "genetic diseases" or the services are termed "genetic services".²

Thus, while public health has had links with genetics over the past 50 years, only in recent years have the terms 'public health genetics' and 'community genetics' been used consistently to refer to the interface between public health and genetics. Given the rapid developments and their broad impact, it is time to review the surveillance of genetic disorders in Victoria.

This article summarises some results from the first stage of an enhanced genetics surveillance system in Victoria. It offers some basic information on the prevalence of selected disorders and on the testing activity related to those disorders. In doing so, it demonstrates a new capacity in Victoria to demystify information made available about genetic conditions. A selection of find-

ings from a detailed report is provided to highlight the public health issues revealed within the data. The report entitled 'Beyond the Crystal Ball: The Epidemiology of Some Genetic Conditions in Victoria' is nearing completion and is expected to be available in hard copy and on the Public Health web site late April 2002.

METHOD

There has been no comprehensive approach to the surveillance of information about genetics in Victoria. In the abovementioned report, we set out to collect data from many sources to present a picture of current status and recent trends, and to ascertain the gaps that need to be filled to establish a comprehensive genetics surveillance system.

Information was collected from the testing laboratories at the major paediatric hospitals and the Birth Defects Register, Perinatal Data Collection Unit, which actively collects information from laboratories, specialised clinics and hospitals on all infants born in Victoria with birth defects from 1982.³ Further information came from the annual *Report on Prenatal Diagnostic Testing*, which is produced by the Perinatal Data Collection Unit and the Murdoch Childrens Research Institute and has information from both public and private testing laboratories from the early 1980s.⁴ Other sources include the Victorian Admitted Episodes Data and Health Insurance Commission data.

RESULTS

The newborn screening program

The only population-based genetic screening program is the Newborn Screening Program, which screens over 60,000 babies born in Victoria every year. Screening started in 1966 with phenylketonuria (PKU). Screening for congenital hypothyroidism (CH) and CF began in 1977 and 1989 respectively, but gene testing started only in 1990. Figure 1 shows the cases identified by the program over the past ten years.

Table 1 shows the estimated birth prevalence, the total prevalence and the comparison with published sources of data for some genetic conditions in Victoria. The birth prevalence of beta thalassemia is estimated to be less than 1 in 100,000 births.

Prenatal testing

Extensive surveillance also occurs as part of the prenatal investigations for conditions such as thalassemia, neural

tube defects and chromosomal anomalies. Figure 2 shows the age distribution of the 163 patients with thalassemia who are currently having regular transfusions at the State-wide thalassemia services at Monash Medical Centre. These are mainly patients with beta thalassemia major.

Figure 3 shows the prevalence of neural tube defects in Victoria for the period 1994-99. Neural tube defects include anencephaly, spina bifida and encephalocele. While there has been a slight increase in their detection due to an increase in the use of maternal serum screening and ultrasound, the prevalence of these defects essentially remained steady from 1983 to 1997, followed by a marked drop in 1998 and 1999.^{6,7}

DISCUSSION

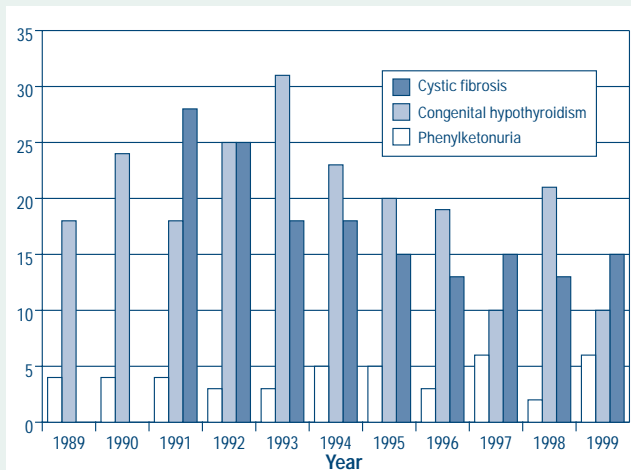
The Newborn Screening Program is a classic public health success story. In two of the three conditions, a relatively simple intervention—namely, the provision of a phenylalanine-free diet in the case of PKU and the provision of a thyroid hormone in the case of CH—reduces the risk of the development of an intellectual disability.

Prenatal diagnosis for thalassemia by cord blood analysis started in 1975, followed by DNA analysis of chorionic villus sampling in 1985. The population distribution of individuals with thalassemia needing transfusions shows the effects of one of the longest running surveillance programs (Figure 2). For the age group 20-40 years old, a higher percentage of affected individuals were born before the introduction of the program and the better possibility of survival with improving treatment. In the past 20 years, only 14 children with beta thalassemia major were born in Victoria.

Table 1: Prevalence of Some Genetic Disorders

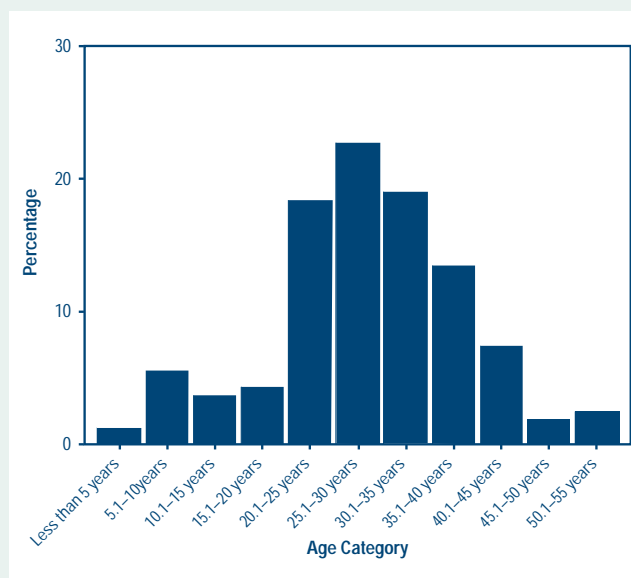
	Birth prevalence	Prevalence	Reported prevalence ⁵
Phenylketonuria (PKU)	1 in 15,700 births		1 in 10,000
Congenital hypothyroidism (CH)	1 in 3,224 births		1 in 3,500
Cystic fibrosis (CF)	1 in 2,900 (1992-95) 1 in 4,500 (1996-99)		1 in 2,500
Neural tube defects	1 in 1,000	1 in 640	1 in 500 pregnancies
Chromosomal disorders	1 in 350	1 in 203	
Down syndrome	1 in 900	1 in 485 (1994-98)	1 in 660
Turner syndrome	1 in 7,900	1 in 3,160 (1994-98)	1 in 2,000 females

Figure 1: Newborn Screening Program's Diagnosed Cases, Victoria, 1989-99



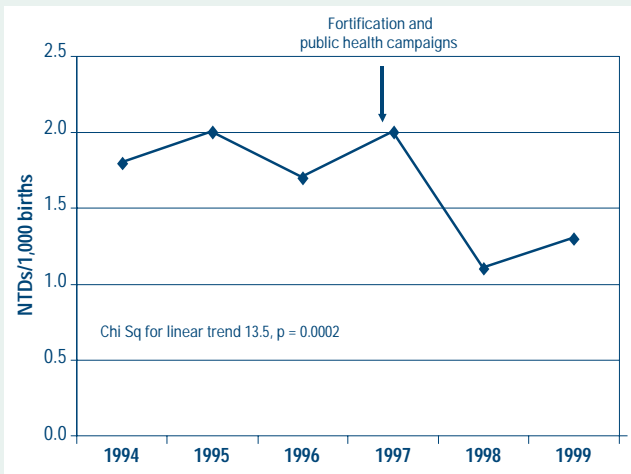
Source: Newborn Screening Program.

Figure 2: Thalassemia—Age Distribution of Patients Being Treated with Regular Transfusions, Victoria, 2001



Source: Thalassemia Services Victoria, Monash Medical Centre

Figure 3: Neural Tube Defects and Folate Intake, Victoria



Source: Birth Defects Register.

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One success story of birth defects surveillance systems was the initial demonstration of an association between neural tube defects and folate intake,⁸ the demonstration of the links with anti-convulsants,^{6,9} and ultimately the demonstration that the campaigns encouraging the use of folate supplements by women before and during the first trimester of pregnancy has worked.^{6,7}

These data must be interpreted with some caution, because there are no published surveillance case definitions for these genetic conditions. Case ascertainment may vary within and between conditions.

CONCLUSIONS

Decisions about targeted screening or population-based screening programs need to be based on the best available data. These surveillance data, along with input from clinical and public health experts and health economists, will inform proper policy analysis. It is important to take a consistent national approach, implemented by all States and Territories. Such an approach has been lacking, but the National Public Health Partnership's appointment of a National Public Health Genetics Working Group is a step in the right direction. Improved surveillance of genetic conditions will be an essential task of the strategy and will contribute to the formulation of good public health policy.

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Christine Stone Grad Dip Epid Biostat MPH MHSc(PHP) works in the Prevention and National Health Priorities team of the Disease Control and Research Unit, Public Health Branch, Department of Human Services. Rosemary Lester MBBS MPH MS(Epid) FaFPHM is Manager of the Prevention and National Health Priorities team.

Contact: Christine Stone—Tel. 03 9637 4196, Fax 03 9637 4653, Email christine.stone@dhs.vic.gov.au

Hepatitis C: Enhancing Routine Surveillance In Victoria

Dr Sean Tobin

ABSTRACT

Hepatitis C is a serious blood-borne infection of epidemic proportions that is proving difficult to control in Australia. Many people acquire and carry the hepatitis C virus without symptoms, so infection surveillance is challenging. The notifiable diseases system is unlikely to ever represent a comprehensive surveillance system for hepatitis C in Victoria. It has the potential, however, to play an important part in the collection of information about the incidence of, and risk factors for, infection. This information is vital to the planning and evaluation of prevention and treatment strategies.

This paper reviews the epidemiology of hepatitis C notifications in Victoria from 1997 to 2000 and outlines Department of Human Services strategies to enhance the notifiable disease surveillance system for hepatitis C infections from 2001.

BACKGROUND

The most current modelling of hepatitis C virus (HCV) infection in Australia estimated that around 190,000 people were living with the infection in 1997.¹ This modelling also estimated that there would be approximately 11,000 acute infections each year, with 91 per cent of cases exposed through injecting drugs.