
CLINICAL AND COMMUNITY STUDIES
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ÉTUDES CLINIQUES ET COMMUNAUTAIRES

The Alberta Hereditary Diseases Program: a regional model for delivery of genetic services

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Genetic counselling and related services are generally provided at major university medical centres because they are very specialized. The need for rurally based genetic services prompted the inclusion of an outreach program in the Alberta Hereditary Diseases Program (AHDP), which was established in 1979; the AHDP was designed to provide services to the entire province through two regional centres and seven outreach clinics. There is a community health nurse in almost every health unit whose duties are either totally or partially devoted to the AHDP; thus, genetic help and information are as close as a rural health unit. The AHDP is designed to provide complete clinical (diagnostic, counselling and some management) services and laboratory (cytogenetic, biochemical and molecular) services for genetic disorders. In addition, the program emphasizes education and publishes a quarterly bulletin, which is sent free of charge to all physicians, hospitals, public health units, social service units, major radio and television stations, newspapers and public libraries and to selected individuals and groups in Alberta.

Étant donné leur niveau de spécialité, les conseils relatifs à la génétique et les services qui s'y rattachent sont habituellement fournis dans les grands centres médicaux universitaires. Le besoin de disposer de services génétiques en milieu rural a donné lieu à la mise sur pied d'un programme décentralisé dans le cadre du programme sur les maladies héréditaires de l'Alberta, le "Alberta Hereditary Diseases Program" (AHDP), créé en 1979. Le AHDP a pour objectif de dispenser des services dans l'ensemble de la province par l'intermédiaire de deux centres régionaux et de sept cliniques décentralisées. Dans la plupart des centres de santé on retrouve une infirmière communautaire qui se consacre en partie ou entièrement au AHDP. Dès lors, tout le monde peut bénéficier d'une aide ou de renseignements en génétique, sans devoir aller au-delà du centre de santé rural. L'AHDP a été conçu de sorte à assurer la prestation de services cliniques complets (diagnostics, conseils et certains éléments de gestion) ainsi que de services de laboratoire (analyses cytogénétique, biochimique et moléculaire) pour des désordres génétiques. En outre, le programme met l'accent sur l'éducation, et un bulletin trimestriel est envoyé gratuitement à tous les médecins ainsi qu'aux hôpitaux, unités de santé publique, centres de services sociaux, stations de radio et de télévision, journaux et bibliothèques publiques et à certains particuliers et groupements de l'Alberta.

Genetic services, which began as counselling, have usually been started by geneticists. Although the frequency of genetic diseases has probably not changed, the relative frequency has increased because of the decline in the incidence of infection and nutrition disorders. Genetic services

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now encompass a much broader concept and include clinical diagnosis, syndrome identification, treatment and management, and specialized laboratory services such as cytogenetics, biochemistry and molecular genetics.

Most genetic services are situated in major university medical centres; some community hospitals also have a genetics program. Therefore, such services have often been unavailable to more rurally based families. In the United States regional or outreach programs were introduced more than a decade ago.¹⁻⁴ A review of genetic counselling re-

sources in Canada was published in 1974 and 1978.^{5,6} Since then outreach programs have been established in British Columbia, Alberta, Saskatchewan, Ontario and Nova Scotia.

We believe that the Alberta Hereditary Diseases Program (AHDP), established in 1979, is unique in its scope and in the extent of its collaboration between hospital-based services and public health services. The AHDP has three main components: (a) the regional centre program, (b) the outreach program and (c) education.

Regional centre program

Alberta has a population of approximately 2.4 million people and 44 000 births annually; these figures are divided almost equally by the northern boundary of the Red Deer Health Unit (Fig. 1). The province is divided geographically into northern and southern regions. Edmonton is the headquarters of the northern region and is responsible for the population in the areas north of the Red Deer Health Unit. Calgary, the headquarters of the southern region, is responsible for the population of the Red Deer Health Unit and all areas south of it. Each regional centre is based in the Department of Paediatrics at the respective university; the current complement of approved positions is provided in Table 1.

The 1988-89 global budget for each centre is approximately \$1.5 million and is administered by University of Alberta Hospital (Edmonton) and Alberta Children's Hospital (Calgary). The budget is protected from the general hospital and university budgets and pays for the entire operation of the program (i.e., salaries, benefits and operating costs).

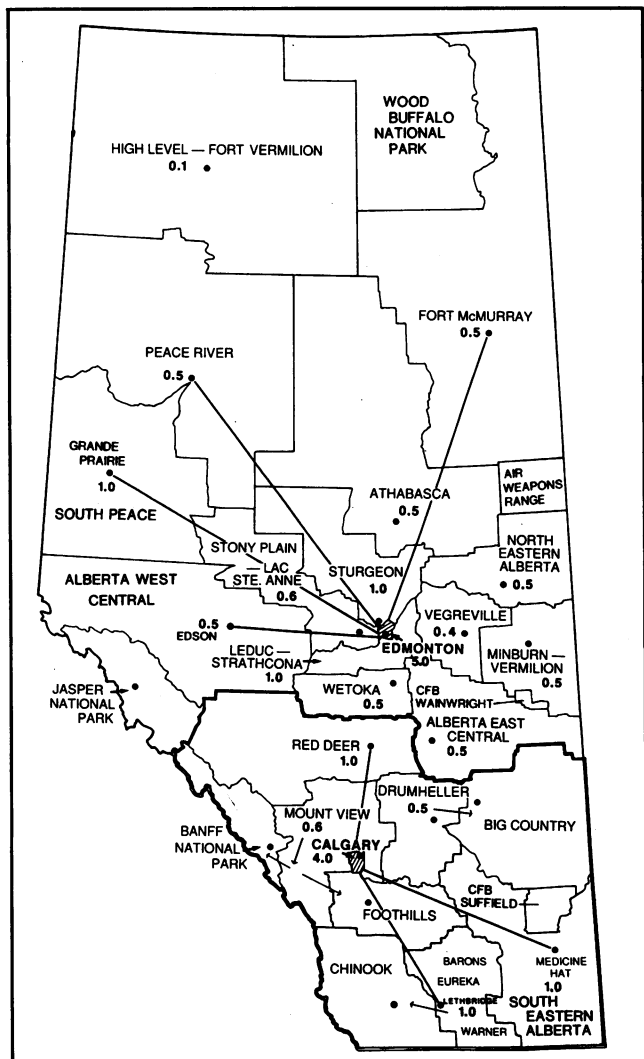


Fig. 1: Map of Alberta, showing the boundaries of the health units and the number of full-time-equivalent nursing positions associated with the Alberta Hereditary Diseases Program. There are four positions in each of the two regional centres; the additional position in Edmonton is part of the public health unit. The double-line boundary divides the northern and southern regions. The heavy lines radiating from the regional centres point to the outreach clinics, and the small arrows indicate areas in which a community health nurse is responsible for more than one health unit. The black dots represent health unit headquarters. CFB = Canadian Forces Base.

Table 1: Number of staff with the Alberta Hereditary Diseases Program (AHDP) in each regional centre in 1988

Position	No. of people
Genetics associate or nurse	4
Clinical geneticist	3
Biochemical geneticist	1
Cytogeneticist	1
Molecular geneticist*	1
Neurologist-geneticist*	1
Ophthalmologist-geneticist†	1
Fellow	1
Health educator*	1
Cytogenetics technologist	6-8
Biochemical technologist	4-6
Molecular genetics technologist*	4
Secretary	5
Other support personnel	2

*Calgary only.

†Person based in Edmonton but conducts clinics in Calgary.

A service molecular genetics laboratory was established in Calgary in 1982 with funds from the Alberta Children's Hospital Foundation; most of its operating costs are now incorporated into the budget for Calgary's hereditary diseases program.

Most of the people requesting the services of the AHDP have been children and their parents; however, there is an increasing demand and need by adults, who are also seen at the pediatric clinics. Antenatal genetics clinics are also conducted in conjunction with each university's departments of Obstetrics and Gynaecology and of Radiology. Ward consultations are done not only at the regional centre hospitals but also at other hospitals in Edmonton and Calgary, particularly in the newborn intensive care nurseries. Ophthalmologic genetics clinics are held in Edmonton and Calgary and neurogenetics clinics in Calgary.

The caseload statistics for 1984-88 for the clinical and cytogenetics components of the AHDP are provided in Table 2. The most marked increases were in services for antenatal diagnosis; however, only 58% of these patients had previously received counselling in our program. In Alberta the proportion of births among women aged 35 years or more has been increasing; the figure was 2367 (5.4% of total births) for 1985, 2486 (5.8%) for 1986 and 2687 (6.4%) for 1987. In 1986-87 a study showed that only 35% of the women in this age group were undergoing antenatal diagnosis; there was a large disparity between the figures from Calgary and those from rural areas in southern Alberta (44% v. 15%).⁷ Likely the biggest expansion in genetic services will be in antenatal diagnosis because of an increase in knowledge and acceptance of testing, an ability to diagnose more diseases and an increase in the number of women 35 years of age or older.

A teratogen service is offered whereby physi-

cians or their patients may obtain advice over the telephone on the genetic and reproductive consequences of exposure to such environmental substances as drugs, radiation and infection. A report is sent to the physician in each instance. We do not have enough staff to do a formal follow-up on each case; however, we are almost certain that we will be notified of any unfavourable outcome.

Outreach program

Outreach clinics are held in Red Deer, Medicine Hat, Lethbridge, Edson, Peace River, Grande Prairie and Fort McMurray by one of the regional centre clinical geneticists. Their frequency depends on local need and for many areas is monthly. Outreach clinics are held in the local health unit, in which examination and interview rooms are provided. A regular clinic is also held in Red Deer at the Michener Centre, a 1400-bed residential facility for the mentally handicapped. Travel to Red Deer and Edson is by car and to the other centres by air.

There is a community health nurse in 26 of the 27 health units whose duties are either totally or partially devoted to the AHDP (some nurses cover more than 1 health unit). The nurse secures all necessary documents, takes the family history, draws up the pedigree, prepares the families for the clinic and is responsible for the follow-up, ensuring that the patient or parents have understood the information discussed. If laboratory investigation is required she is responsible for coordinating the testing at the local hospital and the transportation of specimens to the regional centres. About six to eight families are seen each day at the outreach clinics. This allows for about 1½ hours for each new case and 30 to 45 minutes for each follow-up case. Amniocentesis and

Table 2: Clinical and cytogenetic statistics from the AHDP, 1984-88

Service	Year; no. of patients				
	1984	1985	1986	1987	1988
Clinical					
New patients	577	755	898	936	857*
Antenatal	408	459	554	760	878
Ward consultation	301	346	409	357	374
Review or follow-up	261	328	462	507	602
Outreach clinic	238	235	303	304	300
Total	1785	2123	2626	2864	3011
Cytogenetic					
Leukocyte or fibroblast	1561	1536	1684	1488†	1524
Antenatal	830	907	1124	1188	1511
Hematologic disorder or cancer	112	148	192	191	247
Total	2503	2591	3000	2867	3282

*Numbers down because of death of one clinical geneticist and resignation of another. Could not be replaced quickly.

†Numbers down because of change of policy to examine parental chromosomes only after three or more spontaneous abortions instead of two or more.

chorionic villus sampling are performed mostly at the regional centres; the procedure is sometimes done locally and the fluid sent by courier to one of the regional centres.

Most of the AHDP nurses (at both the regional centres and the outreach clinics) have a bachelor's degree in nursing, which sometimes includes an undergraduate course in genetics. The nurses who have had no exposure to human genetics are encouraged to take a correspondence course organized by Athabasca University, Athabasca, Alta.

The present budget of the outreach program is approximately \$500 000 per year, which covers such costs as the salaries of 13.2 full-time-equivalent positions, travel, education and office supplies.

Education

Education is an important part of the AHDP. All of the clinical and laboratory geneticists at the regional centres have full-time faculty appointments at the respective universities and are thus heavily involved in teaching. Many of the residents and fellows in neonatology at each centre rotate through the genetics division, as do residents and fellows in other departments (e.g., obstetrics, family practice and occasionally internal medicine). The genetics faculty is, of course, involved in the education of practising physicians through speaking at grand rounds and seminars. Talks are also given to the physicians at the outreach clinics.

Education is provided to other groups such as public health unit personnel, hospital nurses, social service workers, students in junior college and in junior and senior high school, and disease support groups. The regional centre in Calgary has a full-time health educator, and the one in Edmonton has an AHDP nurse on the city's Board of Health whose duties are largely educational. The health educator in Calgary is also the managing editor of a quarterly publication (*Bulletin of the Hereditary Diseases Program of Alberta*), which is sent free of charge to all physicians, hospitals, public health units, social service units, major radio and television stations, newspapers and public libraries and to selected individuals and groups.

A large part of the nurses' responsibilities at outreach clinics is the provision of educational services to other community health nurses, hospital nurses, school board members, teachers, social service workers and the community in their area.

Hereditary metabolic diseases

Province-wide screening for phenylketonuria started in 1967 at University of Alberta Hospital, Edmonton.⁸ In 1975 this program was modified to

adapt Scriver's technique for qualitative amino acid identification.⁹ Screening for congenital hypothyroidism began in 1977 with the use of a dried blood spot, identical to the technique used in amino acid identification. Further changes, introduced in 1985, included determining the serum level of thyroid stimulating hormone instead of thyroxine as an initial thyroid test, using an unmeasured blood sample on filter paper, and improving long-term monitoring and follow-up of children with positive results.¹⁰ Centralized follow-up clinics are now held at the two regional centres, each having facilities for definitive diagnosis and initial follow-up therapy. Families are seen for genetic counselling at either a regional centre or an outreach clinic. The mother would have to travel to a regional centre if prenatal diagnosis were required, although the counselling could take place at an outreach clinic.

The number of patients with hereditary metabolic diseases has been small since there are only 44 000 live births per year in Alberta. From 1980 to 1985 the frequency of phenylketonuria and congenital hypothyroidism was 1/16 564 and 1/6464 respectively (Dr. Patrick Ferreira: personal communication, 1989). However, the increased ability to recognize metabolic diseases in the neonate has resulted in many more cases being diagnosed than had been expected.

Advisory committee

The AHDP has an advisory committee that reports directly to the Alberta minister of health. It consists of a chairman appointed on the recommendation of the deans of the two medical schools, representatives from University of Alberta Hospital and Alberta Children's Hospital, the public health sections of the ministry, the Alberta Medical Association, the Society of Medical Officers of Health, the Alberta Community Health Nursing Society and the administration departments of University of Alberta Hospital and Alberta Children's Hospital, the two directors of the regional genetics centres and three lay members. The committee meets three to four times each year to discuss the budget, plans, policies and other critical issues.

Discussion

We believe that the AHDP could be adapted for use in the other provinces or, indeed, in other countries. Riccardi¹¹ provided a detailed description of how to set up an outreach program. Although the AHDP is by no means original we believe it is unique because it gives complete coverage to the entire population through an extensive network of community health nurses responsible to the pro-

gram. Although the primary thrust of genetics has been in the pediatric and obstetric populations the advent of more sophisticated molecular diagnostic techniques will result in a need for more geneticists primarily responsible for adult patients.

Although the AHDP appears to be an expensive program (\$3.5 million for a population of 2.4 million) the prevention of only a few cases of serious hereditary disease such as Duchenne's muscular dystrophy will in time be cost saving and thus lower another section of the provincial government's health budget.

Genetic services are an integral part of modern medicine not only for prevention but also for diagnosis and management. A knowledge of genetics enables many couples to plan families when they would not have otherwise, or vice versa, and it often helps children and their parents come to terms with a hereditary disease or a congenital anomaly. Education is extremely important because genetics will continue to have a significant role in health care.

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Conferences

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June 12-15, 1990: Canadian Hospital Association Annual Conference: Caring for Medicare
Centennial Auditorium and Holiday Inn, Saskatoon
Conferences, Canadian Hospital Association, 100-17 York St., Ottawa, Ont. K1N 9J6; (613) 238-8005,
FAX (613) 238-6924

June 15-19, 1990: Canadian Anaesthetists' Society
47th Annual Meeting
Hyatt Regency Hotel, Vancouver
Ann Andrews, executive director, Canadian Anaesthetists' Society, 187 Gerrard St. E, Toronto, Ont. M5A 2E5;
(416) 923-1449

June 24-27, 1990: Canadian Nurses Association Annual Meeting and Biennial Convention
Calgary Convention Centre
Linda O'Rourke, corporate affairs manager, Canadian Nurses Association, 50 The Driveway, Ottawa, Ont.
K2P 1E2; (613) 237-2133, FAX (613) 237-3520

June 24-27, 1990: Canadian Society of Otolaryngology
44th Annual General Meeting — Head & Neck Surgery
Bonaventure Hotel, Montreal
Canadian Society of Otolaryngology — Head & Neck Surgery, 103-4953 Dundas St. W, Islington, Ont. M9A 1B6; (416) 233-6034, FAX (416) 239-8220

Aug. 27-29, 1990: Canadian Health Economics Research Association 4th Conference: Restructuring the Health Services System — How Do We Get There from Here?
University of Toronto
Gail Thompson, conference coordinator, Institute of Health Management, University of Toronto, 12 Queen's Park Cres. W, Toronto, Ont. M5S 1A8, (416) 978-8384, FAX (416) 978-7350; or Dr. Raisa Deber, conference chair, Department of Health Administration, University of Toronto, (416) 978-8366

Sept. 13-15, 1990: New Brunswick Medical Society
Annual General Meeting
Hotel Beauséjour, Moncton
Ms. Judy Orem, annual general meeting coordinator, New Brunswick Medical Society, 176 York St., Fredericton, NB E3B 3N8; (506) 458-8860

Oct. 1-5, 1990: Canadian Society of Forensic Science
Annual Conference
Skyline Hotel, Ottawa
Abstract deadline is June 1, 1990.
Canadian Society of Forensic Science, 215-2660 Southvale Cres., Ottawa, Ont. K1B 4W5;
(613) 731-2096