ATLANTIC RESEARCH CENTRE FOR MENTAL RETARDATION

PROGRESS REPORT JULY, 1968 to JUNE, 1969.

Since the last Board meeting, approximately one year ago, much has been accomplished through the expanded activities of the Atlantic Research Centre.

In August, 1968, the scientific group was greatly strengthened by the arrival of our newly appointed population geneticist, Dr. Richard F. Shaw. Dr. Shaw came to us from Wayne State University where he had been Assistant Professor of Biology and Associate in Pediatrics since 1965, having obtained his Ph.D. earlier from the University of California. He is author of a number of significant publications in the field of population genetics, and has a particular interest in the application of the computer to the solution of problems in population genetics.

Mr. Allen Smith, a computer programmer with experience in genetics, was appointed shortly after Dr. Shaw's arrival. Mr. Smith had previously been working in the cytogenetics laboratory of The Children's Hospital in Detroit.

Dr. Shaw has been adapting his computer methods for population genetics to the Dalhousie computer, and will seek applications of these methods to available data in the field of mental retardation.

During the past year, the Board approved support of a senior research technician to assist Dr. M. DeWolfe with her

investigative work. This appointment was delayed unavoidably by Dr. DeWolfe's illness, but a candidate has now been selected, and we expect the appointment to be made in the very near future.

We continue to study families with a variety of genetic problems and to offer counselling service to such families and to the medical community, both local and distant. Since March 1968, 150 Chromosome analyses have been carried out and approximately 40 families have been studied. One of the families mentioned in the last progress report has now been investigated thoroughly and was reported at the annual meeting of the Canadian Paediatric Society. A noteworthy feature in this kindred was the occurrence of a dermatoglyphic pattern more commonly associated with mongolism. Three families in which a child has been born with the Cri-du-Chat syndrome have been studied during the past year. In one the father was found to be a mosaic (i.e. showing more than one genetically distinct cell line) for the same cell line as that seen in his affected child. This finding permitted meaningful genetic counselling to be offered to the family. This phenomenon will be the subject of a future publication, since this occurrence has never been reported previously. The syndrome is associated with severe mental retardation and the finding of this condition in the two other children mentioned has been of considerable prognostic value.

Another major project currently under way involves a survey of all Halifax school children by height and weight. The purpose of this investigation is two-fold: First, to obtain normative data on development; second, to enable us to undertake cytogenetic screening of certain sub-sets of this population for chromosome abnormalities. The importance of ascertaining such individuals, particularly males with an extra Y chromosome, in the normal population, with a view to observing their mental and social development, can hardly be over stressed. Some 10,000 children have now been examined by height and weight and the data is being processed by computer. Chromosome analysis has already been carried out on a certain number of blood samples from the "high risk" sub-groups of this population. A provisional report of this survey has already been accepted for presentation at the Third International Conference on Congenital Malformations to be held in September at the Hague.

Dr. Welch has also continued his investigations of Downs Syndrome and the XYY syndrome. Some results of these studies have been published or presented by Dr. Welch as an invited speaker to the First Conference on the Clinical Delineation of Birth Defects held at Baltimore in May 1968; by invitation at the Centennial Symposium of the Faculty of Medicine at Dalhousie in September 1968; and as an invited participant in a seminar sponsored by Howard University in Washington in March 1969. All three papers will be published and a more

detailed report of these investigations will be published subsequently.

With respect to the survey for individuals with the XYY anomaly among recidivists of the Atlantic Provinces, further preparatory work has been done through visits to Dorchester Penitentiary. Permission has been obtained from Federal Authorities for the full survey to be undertaken.

Some progress has been made in investigating the variant of Niemann-Pick disease occurring in the French Canadian group living in the vicinity of Yarmouth, Nova Scotia. However, travelling difficulties have proved to be a considerable handicap in view of the limitations of staff, and this endeavor has not proceeded as fast as had been anticipated.

Two other investigations should also be mentioned. One concerns a kindred in whom three distinct genetic diseases appear to be segregating, one of them being an apparently "new" genetic condition associated with severe mental retardation which will be the subject of a future publication. The second concerns a group of girls found to have coarctation of the aorta, this being a known complication of Turner's syndrome. The investigation involved a search at both clinical and cytogenetic levels for evidence of minor signs of Turner's syndrome. The results indicated that some children in this group do have cytogenetic anomalies; these results were communicated in a paper presented by Dr. Welch at the

Annual Meeting of the Canadian Society for Clinical Investigation in Vancouver.

At the time of writing this report we are sorry to say that we are about to lose one of our more experienced technicians Mrs. L.Y. Lee; who is moving with her husband to Liverpool, England. We are happy to add that she proposes to continue work in cytogenetics. She intends to work towards a higher qualification and with this end in view she has been awarded a bursary by the Canadian Association for Retarded Children. Two further important scientific appointments to the Atlantic Centre have been made and are expected to add great strength to the research program. Matthew Spence, M.D., Ph.D. is a brilliant young neurochemist, currently Assistant Professor of Experimental Medicine at McGill University and conducting research in brain chemistry in the laboratories of the University Clinic at The Montreal General Hospital. He is a Medical Research Council Scholar and holds important research project grants from the Medical Research Council of Canada. Dr. Spence has accepted an offer of an appointment as Associate Professor of Paediatrics at Dalhousie with additional appointments of Assistant Professor of Biochemistry and Lecturer in Psychiatry. He expects to join our staff later this year and will occupy the laboratories presently assigned to Dr. DeWolfe, when she transfers her activities to the new laboratories in the Izaak Walton Killam Hospital for Children.

His principal responsibility will be the conduct of research in neurochemistry in the Atlantic Centre laboratories. In addition, following upon an agreement between the Departments of Paediatrics and Psychiatry, he will be responsible for the development of an integrated program of neurochemical research involving both departments and including the laboratories contiguous to those of the Atlantic Centre. Coupled with the research activity, we expect to establish a Ph.D. training program in neurochemistry.

The second appointment is that of Dr. Margaret Corey. Dr. Corey is a Ph.D. cytogeneticist, currently holding an appointment as Assistant Professor of Paediatrics at the University of British Columbia and conducting research in the Division of Medical Genetics. Like Dr. Spence, she is a Medical Research Council Scholar, recipient of a valuable research project grant from the MRC, and author of several important publications in cytogenetic research. She expects to join our staff in August or September. Negotiations are currently in progress for the transfer of both MRC scholarships to Dalhousie. Once this is achieved, it is expected that their research grants will also be transferred.

With these two further appointments the basic scientific staff of the Centre will be complete and all available laboratory space will be in full use. If progress to date is indicative of future prospects we should look forward to exciting achievements during the next few years.

DALHOUSIE UNIVERSITY
HALIFAX, N. S.

BUSINESS OFFICE

Atlantic Research Centre for Mental Retardation

Dalhousie account - X-20-1

Receipts:

Unexpended balance at May 31, 1968 (per statement)		$30,696.60	
December 30, 1968, Province of Prince Edward Island		3,150.00	
April 8, 1969, Province of Nova Scotia		18,000.00	$51,846.60

Expenditures:

Salaries:			
Dr. J. P. Welch, portion of Pension Fund, Group Insurance and LTD not paid by Dept. of National Health and Welfare grant-			
- quarter ending June 30, 1968	$ 136.44		
- quarter ending September 30, 1968	146.71		
- quarter ending December 31, 1968	146.59		
Others, as above	27.46		
Office overload	157.58		
Allen P. Smith, March, April, & May, 1969	2,045.70		
Susan Hong, May 5-31, 1969	275.00	2,935.48	
Travel:			
Dr. S. Abraham	50.50		
Dr. J. P. Welch	856.90		
Dr. L. K. McNeill	32.00		
Dr. M. W. Spence	114.92	1,054.32	
Other:			
Catering services	153.15		
Allen P. Smith, Computer services	400.00		
Equipment- IBM	335.90		
Exchange on P. E. I. Cheque	1.95		
Blood Collections	11.00	902.00	4,891.80
Unexpended balance at May 31, 1969			$46,954.80

Prepared by:

R. M. Walters
Research Accountant

PUBLICATIONS BY MEMBERS OF THE ATLANTIC RESEARCH CENTRE FOR MENTAL RETARDATION, 1968-69.

Shaw, R.F. and Dreifuss, F.E. Mild and severe forms of X-linked muscular dystrophy. Arch. Neurol. 20:451, 1969.

Shaw, R.F. Cystic fibrosis and genes in populations. Can. Med. Assoc. Jour. (in press).

Welch, J.P. Down's Syndrome and human behaviour. Nature, 219:506, 1968.

Welch, J.P., Clower, C.G. and Schimbe, R.N. The "pink spot" in schizophrenics and its absence in homocystinurics. Brit. J. Psychiatry 115:163, 1969.

ADDRESSES

Dr. R.F. Shaw - "Genetic Screening of the Newborn" American College of Gynecology and Obstetrics.'

"Population Genetics and Health" - Centennial Symposium, Faculty of Medicine, Dalhousie University, 1968. (to be published)

Dr. J.P. Welch - "The XYY Syndrome" - presented at the Centennial Symposium, Faculty of Medicine, Dalhousie University, 1968. (to be published)

"A new syndrome of inherited skeletal dysplasia - report of a family". Canadian Paediatric Society Annual Meeting, 1968.

"Investigations in females with coarctation of the aorta". Canadian Society for Clinical Investigation, Annual Meeting, 1969.