

THE UNIVERSITY OF MANITOBA

A PRELIMINARY INVESTIGATION OF CONGENITAL HIP DISEASE  
IN THE ISLAND LAKE RESERVE POPULATION, MANITOBA

by

Joan Marion Walker

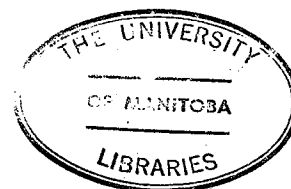
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## Chapter 1

### INTRODUCTION

The study of populations is basic to anthropology, epidemiology, and genetics. Population studies focus attention on groups rather than the individual, and permit study of statics and dynamics relative to evolutionary processes.<sup>101</sup>

Epidemiology requires accurate enumeration of the population at risk of the disease. An advantage of population studies to biological and medical science lies not only in understanding the genetic basis of variability, but also in elucidating the environmental determinants of diseases.<sup>151,4</sup>

Definition of population differences in disease susceptibility is permitted. Differences in disease incidence between populations can be described in demographic and biological terms, making the study of differences in environmental determinants more effective.<sup>151</sup>

Congenital malformations are, increasingly, important foci for epidemiologic and genetic research. Their relative incidence has increased, due to decrease in mortality from other conditions and partly due to improved diagnosis.

Defects of bone and joint systems are the most frequent in

many studies.<sup>108,222</sup> The hip joint is more often involved when dislocation occurs, in isolation or as part of a 'complex' of congenital abnormalities.

The majority of common malformations are considered to be of multifactorial causation.<sup>34,123</sup> Smithells<sup>195</sup> noted that an epidemiological approach is therefore likely to be the most fruitful in the long run. Neel<sup>145</sup> suggested integration of epidemiologic-genetic strategies in research of congenital malformations and chronic diseases.

#### Island Lake Research Project

Corrigan and Segal<sup>47</sup> reported an estimated prevalence of 60 cases per thousand for congenital dislocation of the hip (CDH) in the Cree-Ojibwa Island Lake population. Gray<sup>76</sup> verified the high frequency of CDH. In addition, these authors reported frequent consanguinity in the Island Lake population, and associated CDH with consanguineous marriage. CDH incidence of one<sup>232</sup> to four (neonates included<sup>171</sup>), in 1,000 live births can be given for Caucasian populations.

Research commenced in 1968 by Dr. D. Rokala, Department of Anthropology, University of Manitoba, has generated a genetic-demographic data base for the Island Lake reserve population which spans six generations. Relevant demographic data has been integrated into individual records.

Records have in turn been integrated into biological sibships, with individuals referenced into a single sibship sharing a common progenitor and progenitrix. Sibships have been linked into biological lineages. The data base has been compiled from research in archives, vital records registers, church and administrative records, and interviews. Rokala<sup>170</sup> has stated the specific objectives of the research as:

1. to ascertain the segregation frequency, mode of inheritance or heritability, and recurrence risk for congenital hip disease (CHD) and bifid uvula, a reported microform of cleft palate.
2. to investigate the population structure of Indian reserves characterized by elevated prevalence of one or more diseases of purported genetic etiology.

The target population was characterized by high fertility, high mortality, and filter immigration rates following the establishment of the Island Lake reserve with subsequent familial emmigration.<sup>170</sup> In small isolated populations consanguinity may result in elevated prevalence of diseases which involve, at least in part, genetic or familial mechanisms.<sup>184,220,221,218,85,170</sup> Rokala<sup>170</sup> suggested that the Island Lake population might be considered "an isolate with elevated random or non-random consanguinity between mating types as contributory to the population pathology." Schull<sup>185</sup> reported Japanese studies on consanguinity that revealed a picture of small but pervasive effects detectable

only with large numbers of observations.

### Proposed Study

This study is a preliminary stage in the proposed investigation of congenital hip disease (CHD)\*. The target population includes the four administrative bands of Treaty Indians resident at Island and Red Sucker Lakes. Treaty Indians with residence outside this area who have been traced to the Island Lake population due to prior residence and/or genealogical connections are included. Objectives of this study are:

1. Description of basic epidemiological features of CHD. These include sex ratio, unilateral/bilateral hip involvement, left/right hip involvement, seasonality, birth order, birth presentation, birth weight, parental age, prevalence and variability within the atypical/teratological cohort.
2. Calculation of annual prevalence rates. Consideration of the trends in specific subtypes of CHD should enable more definitive statements to be made concerning the future health care needs within the target population.
3. Description of familial clustering. Families 'at risk' may be identified should familial clustering be revealed.
4. Consideration of the attitude of members in the target population, affected and unaffected, to CHD, and the functional disability experienced by probands.

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\* The term congenital hip disease will be employed to encompass the variability in pathological changes, designated specific terms (dysplasia, subluxation, frank dislocation) by medical practitioners.

### Relationship to Total Project

A CHD data base has been created which has been integrated into the genetic-demographic data base, and will permit investigation of the genetic processes involved. Data are based on attempted complete ascertainment of all newborns in the target population since 1955. Representation of all families, not just those exhibiting the disease is therefore possible. Investigation of associated anomalies may detect genetic markers of utility in intra- and inter-population analyses.

### Outline of Methods

Radiological examination of all members in the target population is necessary to elicit complete ascertainment of CHD. This has not been undertaken. Probands have been identified through retrospective study attempting truncate ascertainment, locating probands from medical records. 28,217 Personal interviews and observations were employed to obtain data on attitude and functional disability, and detection of social factors which may have bearing on the incidence of CHD in the target population. Data description has been conducted with the aid of the SPSS (Statistical Package for Social Sciences) system of computer programs.

### Sources of Data

Data on the target population was derived from the following sources:

1. Winnipeg Office, Northern Medical Services, Department of National Health and Welfare (DNHW).
2. Records of medical personnel (Segal, Gray, Singh) who, in the employment of DNHW, conducted annual CDH surveys in the target population, commencing in 1949.
3. Medical records of six Winnipeg hospitals (Children's, General, Shriner's, St. Boniface, Rehabilitation, and the D.A. Steward Centre), and the Charles Camsell Hospital (DNHW), Edmonton.
4. Health station records at the Garden Hill, St. Therese Point, Wasagamach, and Red Sucker Lake communities.
5. Field work of approximately six weeks duration in the four communities of the Island Lake reserve.
6. The genetic-demographic data base was utilized to complete data from all sources and to locate probands in the genealogical matrix.
7. Provincial Atmospheric Environment Canada records were obtained for utilization in the analysis of seasonality.

### Organization of Data

The data was numerically coded and placed on two computer cards per individual. Each individual has a unique identity number, identical to that used in the main data base. Record linkage is based on the individual identity numbers, randomly coded surnames, and birthdates. All information on individuals and families, derived from all sources, is thereby treated in a confidential manner. CHD cards are



sequenced in the main data base.

### Theoretical Support

Edwards<sup>59</sup> noted that the knowledge of pathologic aspects of human population genetics was severely limited by the paucity of data relevant to the genetic structure of human populations. The need for extensive data, to permit investigation of the transmission of hereditary traits and detection of environmental factors is well recorded.<sup>217,218,185,151,25</sup> The data presented herein, are based on annual surveys with attempted complete ascertainment of all newborns. These data are therefore considered representative of the population of individuals born in and from 1955 to the present. The problem of bias inherent in hospital samples is minimized by the majority of probands located from hospital records having been previously ascertained in the annual surveys. Additional sources of bias in ascertainment are reduced as the population is composed of Treaty Indians (with an, as yet undefined, degree of white admixture), with similar environment and socio-economic status.

The epidemiological parameters to be investigated have been delineated in numerous reports and studies on CHD.<sup>218,217,161,232,176,233,50</sup> Definition of these parameters permits inter-population comparison. Detection of similarities or differences between populations may contribute to

the understanding of CHD etiology. Increased incidence amongst relatives is not proof, by itself, that inheritance is involved. It may be due to environmental factors shared by family groups.<sup>158</sup> The continued custom of swaddling infants in cradleboards is one of the environmental factors existent at Island Lake. This factor is implicated in prevention of natural remission of the neonatal dysplastic hip.<sup>176,98</sup>

In the majority of modern industrialized populations acceptance of infant prophylactic and/or remedial treatment precludes study of the natural progression of the disease and functional disability attendant on advancing age. Non-availability of treatment prior to 1949, long standing reluctance and limited acceptance of treatment by members in the target population since 1949, permits investigation of the natural progression of the disease. Comparison is therefore possible with the Navajo, Many Farms study<sup>161</sup> in which similar features were present.

The data base may enable delineation of association between major defects and CHD, a factor Neel<sup>145</sup> noted as frequently absent in studies. Atypical and teratological cases arising from developmental defects early in foetal life are reported but excluded from the analysis proper. These cases are usually clinically distinct and are considered

also to be separate etiologically. 67,213,176

### Thesis Format

A review of the extensive literature on CHD is presented in Chapter 2. Definition of the population of Island Lake reserve within a temporal and spatial context is presented in Chapter 3. Historical aspects of medical services in the area, and the CDH project (DNHW) are included to provide background information from which the CHD data base originated. The fourth chapter contains data presentation, epidemiological features, functional disability assessment, and description of cradleboard usage.

Discussion of data, hypotheses, and indications for future health care needs form the major portion of the fifth and final chapter.

## Chapter 2

## REVIEW OF THE LITERATURE

Congenital dislocation of the hip has been known to man since Hippocrates (460-357 B.C.) who first described and named the condition characterized by 'a waddling gait'. Roser (1879) recognised that diagnosis could be made soon after birth, and Ortolani (1937) described a test to detect dislocation in children under one year of age. Routine screening of neonatal hips was not commenced until the early 1950's.

Prior to routine neonatal examinations, the incidence rates reported for CDH were approximately 0.65 per 1,000 live births<sup>163</sup> to 1.0 per 1,000.<sup>35</sup> With the introduction of routine screening in neonates, and inclusion of neonatal 'unstable' hips, incidence rates have shown a fourfold increase.<sup>227,19,191,91,137</sup>

Definitions

It is doubtful that more than two percent of cases diagnosed CDH are in fact complete frank dislocations.<sup>213</sup> Salter (1968:933) commented that in most congenital

abnormalities:

The anatomical deformity is maximal at birth and it is obvious that the deformity has been present from an early stage of intra-uterine development. In congenital dislocation of the hip, by contrast, the anatomical deformity is minimal at birth and, if untreated, becomes progressively more marked during postnatal growth.

McKeown<sup>129</sup> considered that a general definition of congenital malformations should include 'any microscopic or macroscopic structural abnormality'. This definition is applicable to CDH. Howorth (1963:172) commented, ". . . the word congenital is useful as a date line but it is not inclusive". Synonyms in common usage are 'congenital luxation', 'congenital hip disease', 'pre-luxation', 'dislocatable hip', 'congenital dysplasia', 'acetabular dysplasia', 'instability of the hip', and 'unstable hip'. In Italy, 'pre-dislocation' is preferred to dysplasia.<sup>30</sup>

Tuell (1966: 225f.) has defined the three conditions, dysplasia, subluxation (pre-luxation), and dislocation (luxation) as follows:

Congenital dysplasia of the hip is a condition in which the head of the femur is in full contact with a formed acetabulum that is commonly more shallow than normal, and whose roof is more sloping than normal.

By subluxation is meant a hip with the femoral head riding in the rim of the dysplastic acetabulum.

In CDH no part of the head of femur is in contact with the acetabulum.

The head of femur is always dislocated upwards, the direction

may be anterior, lateral, or posterior. Dunn (1969:1037) considered it rational to define CDH as: "an anomaly of the hip joint, present at birth, in which the head of femur is, or may be, partially or completely dislocated from the acetabulum."

### Classification of Types

There is no universal agreement on the classification of CDH. Hass's 1951 classification is frequently used.

Summarized this is:

1. Typical
  - a. dysplasia
  - b. subluxation
  - c. dislocation
2. Atypical
3. Isolated cases of specific known etiology

Tachdjian<sup>205</sup> distinguished two main groups:

1. Teratologic - develops in utero
  - a. part of a generalized congenital abnormality
  - b. independent
2. Typical - characterized by its postnatal appearance
  - a. the 'unstable hip'
  - b. the 'subluxated hip'
  - c. the 'dislocated hip'

In 1967 Finlay, et al., proposed a classification based on the clinical state of the hip with expected findings for each level. There has been little attempt to adopt this classification. Mitchell (1972:4) used:

1. confirmed positive 'clunk' test, condition classified as luxation
2. unconfirmed 'clunk' test but laxity found, classified as 'unstable'
3. no abnormality found, classified as normal\*

Terms in current usage are those defined by Wynne-Davies (1970:315):

1. Primary
  - a. neonatal CDH: diagnosed in the first four weeks of life, usually in the first week, with a 'clunking hip'
  - b. late-diagnosis CDH: diagnosed after the first four weeks, usually months or years later. A well defined group in which there is invariably deformity and no doubt as to diagnosis.

#### Atypical CDH

'Atypical', 'true', or 'teratological' CDH cases are clinically distinct, and the majority are likely to be separate etiologically. A high incidence of CDH in children with multiple congenital abnormalities has been noted. "In many of these, CDH represents a minor anomaly that is not emphasized" (Warkany, 1971:994). These constitute approximately 2 percent of all cases of CDH.<sup>97</sup> Prevalence rates quoted range between 4.3 percent to sixteen percent.<sup>140,163,158,174,50</sup> Variability exists in reported studies whether teratological cases are excluded from CDH studies.<sup>138,67,44,152</sup> The common associated abnormalities and anomalies in

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\* Italics in the original

in 'atypical' CDH recorded in the literature, are presented in the Appendix, Table 1, p.134. Similarities exist between typical CDH and several of the associated anomalies (joint laxity, plagiocephaly) which are present in index cases and their relatives in greater frequency than would be expected from a random sample of a population.<sup>234</sup> Wynne-Davies (1970:715) considered it possible, ". . . there is some change in the type of collagen or in its rate of maturation."

#### Normal Development

Strayer<sup>203</sup> in a major study of the embryology of the hip joint noted that all the elements differentiate from one mass of the blastema. Rotation of limb buds occurs prior to separation of the component parts of the hip joint.<sup>16</sup> The joint cavity opening commences in the 23mm. specimen. The joint space is visible in the 33mm. specimen, and is completed in the 37mm. specimen, approximately at eight to nine weeks of fertilization age.<sup>202,123,135</sup> Gardner and Gray<sup>70</sup> concluded that processes up to this stage are genetically determined, and that intra-articular structures arise in situ. CDH can not occur before the opening of the joint cavity.<sup>202,135</sup> The role of movement in joint development is emphasized, with functional modelling of the hip joint considered especially marked in the first year of life.<sup>111,</sup>



79,55

Diagnosis\*

Neel<sup>144</sup> commented that CDH is more diagnosable at one year of age. Researchers concur in the diagnostic signs of CDH in late-diagnoses but variability is evident in the diagnostic tests employed and considered valid for the neonate. Two tests commonly used in the neonate or early postnatal infant are Ortolani's sign and Barlow's sign (Salford test). Ortolani's sign was devised for hips over six months of age. Barlow found it unreliable in the newborn and modified it slightly.<sup>191,201</sup> MacKenzie (1972:20) commented:

. . . that the "jerk" or "jolt" described by Ortolani was mistranslated as "click" in English. Fascial clicks in the region of the hip, and vacuum clicks arising in the hip and knee are common in infants and should not be confused with the jerk of an unstable hip.

The 'clicking' hip test is more accurately described as the 'clunking' hip test as the positive sign is a 'heavy sound'. Many of the positive high pitched clicks present in the newborn disappear in a few days.<sup>137</sup>

Hart's test for limitation of abduction in flexion is considered reliable by the majority of examiners. With

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\* Diagnostic determinants are given in Appendix B.

dysplasia and dislocation, abduction is usually limited to between 45 and 50 degrees (normal 90°), and if less than 60 degrees an x-ray is warranted. The rule in the neonate does not preclude CDH later.<sup>83</sup> Lowrie (1970:73) considered that ". . . limitation of abduction alone does not indicate a diagnosis of CDH, reflecting only adductor tone." The test was considered of no value by Barlow in 1962. In series of newborn and older children, unequal abduction between sides was found in a small percentage, at all ages.<sup>175,29</sup>

Babb and Sundberg (1970:15) considered telescoping, the 'push-pull' sign to be ". . . probably the most dependable sign." This is present in roughly four percent of newborns and is rare at twelve months.<sup>175</sup> The sign of asymmetrical skin creases is regarded as unreliable. Ryder and Mellbin<sup>175</sup> noted that only fifty to sixty babies in 1,501 had symmetrical skin folds about the hip. In a series of 139 cases Barlow (1962:295) found:

. . . far less than half the infants with dislocated hips had asymmetrical skin folds, and the great majority of children with asymmetrical skin folds were found to be normal.

There is general agreement on the value of radiographs at three to four months of age and thereafter. They are, however, considered of limited value in the neonate.<sup>62, 211,118</sup> In the newborn the relationship of the femur to the