THE COMBINED USE OF A CENTRAL REGISTRY AND VITAL RECORDS FOR INCIDENCE STUDIES OF CONGENITAL DEFECTS

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Current interest in the aetiology of congenital defects has increased the need for reliable sources of information about incidence. A central registry with access to a wide variety of reporting sources can play an important part, not only in incidence studies, but in the general epidemiology of these defects.

Such a registry provides a storehouse of readily available and constantly expanding information. By virtue of the diversity of information and the large samples available, important incidence variables, such as sex, race, maternal age, or season of birth, can be taken into account without sacrificing statistical validity. Moreover, the examination of bulk data of this kind often leads to the discerning of epidemiological patterns such as frequency of specific combinations of defects and familial tendencies.

The Registry for Handicapped Children and Adults in British Columbia was established in 1952. At the end of 1965, there were about 30,000 live and dead registered cases, including 20,300 living children or roughly 3 per cent. of the child population of the Province. It is emphasized, however, that this caseload cannot be used as a source of complete information, for the following reasons:

- (1) Registration is voluntary.
- (2) Until recently minor defects without residual handicap were not registered.
- (3) An individual must be alive at the time in order to be registered. (However, those who die subsequently remain on the Registry caseload.)

It is for these reasons that Registry data must be supplemented by information from Provincial Vital Records. Fortunately, a unique feature of the Registry is its close working association with the Division of Vital Statistics and other bodies within the Public Health organization of the Province. This aspect of the Registry is discussed by Mott (1963) in his account of its history and operation.

Previously reported findings based on the conjoint use of Provincial Vital Records and Registry data have dealt mainly with factors influencing the incidence of specific defects. Miller (1964) found that the incidence of cleft lip and palate among the Indians of British Columbia was higher than among Caucasians whereas the pattern was reversed in the case of neural tube anomalies. Renwick, Miller, and Paterson (1964) presented evidence showing that maternal age influenced the incidence of congenital heart disease independently of its association with mongolism. Newcombe (1966) studied familial tendencies in children's diseases by computer linkage and found that almost all the conditions he examined tended to repeat in sibs.

The present communication describes ways of making the Registry data most effective as a source of ascertainment, by combination with different kinds of vital records. It will be shown that the relative importance of the Registry or of any one kind of record depends on the nature of the defect in question. Of particular importance in this respect are:

- (a) The relative ease of detection of the defect.
- (b) The relative lethality of the defect in the perinatal period.

AUGMENTATION OF INFORMATION FROM LIVE BIRTH RECORDS BY REGISTRY DATA

The Registry, as a source of incidence data, has two main advantages. The first is access to information relating to defects reported on live birth notices by attending physicians. Through an arrangement with the Division of Vital Statistics, registration of newborn children with defects reported on these records has been in effect since the inception of the Registry. The second advantage is the long period of casefinding inherent in the operation of the Registry, resulting in the subsequent registration of many cases which are "missed" at birth. This advantage is provided by the province-wide reporting of handi-

capped children to the Registry by provincial and metropolitan health units, voluntary agencies, institutions, hospitals and treatment centres, private physicians, and other sources.

Table I shows the number of live-born children reported on the Physician's Notice of Birth in British Columbia in the years 1952-57 inclusive, as having specified congenital defects, compared with the number of live and dead children originating from the same cohort of births, who were on the Registry at the end of 1964. Also shown are incidence rates per 1,000 live births based on data from these two sources. It can be readily seen that live birth notifications alone are unreliable as a source of incidence data for congenital heart disease, and are inadequate even for some of the gross malformations, such as clubfoot and the cleft lip and palate complex.

TABLE I
CASES WITH SPECIFIED DEFECTS FROM THE 1952-57
COHORT OF 203,642 LIVE BIRTHS IN BRITISH COLUMBIA

Malformation	Reported on Live Birth Notifications		On the Live and Dead Registry Caseload End of 1964	
	Number	Rate per 1,000 Live Births	Number	Rate per 1,000 Live Births
Clubfoot	370	1.82	494	2.43
Cleft palate and/or Cleft lip ..	280	1.37	367	1.80
Spina bifida ..	186	0.91	199	0.98
Hydrocephalus ..	48	0.24	163	0.80
Congenital heart disease* ..	78	0.38	794	3.90

* In this paper this term denotes all cardiovascular defects under the general rubric 754 of the International Classification of Diseases (1955 Revision) as a single entity. A case with more than one of these defects is counted only once.

The important point is that for most defects there is a residue of cases which are not reported to the Division of Vital Statistics within the 48 hours required by law for the submission of a Physician's Notice of a Live Birth or Stillbirth Notification. This omission may be due to oversight in the case of overt malformations, or to the essentially occult nature of the condition as in the case of congenital heart disease, or to post-natal development, as in some forms of hydrocephalus.

Since casefinding may extend over a period of many years after birth, the importance of allowing as long a follow-up period as possible cannot be over-emphasized. This is demonstrated in Table II, which shows the number of live and dead cases on the Registry at the end of 1964, who were born in the biennial periods shown, expressed as rates per 1,000 live births in British Columbia in those periods. There is a general decline with decreasing age. The importance of the schooling period in the casefinding of congenital heart diseases is evident from the Table. There is a high plateau from 1952-57. The live children born in these years were between the ages of 7 and 12 years in 1964, while, with the partial exception of the 1958-59 cohort, the remaining cohorts were still in the pre-school age groups.

The advantage of an extended casefinding period does not apply in the case of an extreme type of malformation such as anencephaly, which is inevitably recognized at birth and is confined largely to stillbirths. For such malformations, live and stillbirth notifications used in conjunction are a more adequate source of information than the Registry statistics. For most severe defects, Registry data must be supplemented by information from death and stillbirth records, and the degree to which this is necessary is

TABLE II
BRITISH COLUMBIA-BORN REGISTRY CASES WITH SPECIFIED DEFECTS AT THE END OF 1964
BY PERIOD OF BIRTH*
Showing Numbers and Rates per 1,000 Live Births

Biennial Period	No. of Live Births	Clubfoot		Congenital Heart Disease		Spina Bifida		Hydrocephalus		Cleft Palate and/or Cleft lip	
		No.	Rate	No.	Rate	No.	Rate	No.	Rate	No.	Rate
1952-53	61,573	159	2.58	240	3.90	62	1.01	59	0.96	99	1.61
1954-55	67,084	167	2.49	261	3.89	74	1.10	51	0.76	137	2.04
1956-57	74,985	168	2.24	293	3.91	63	0.84	53	0.71	131	1.75
1958-59	79,548	184	2.31	236	2.97	73	0.92	59	0.74	129	1.62
1960-61	78,707	131	1.66	161	2.05	48	0.61	31	0.39	124	1.58
1962-63	75,606	122	1.61	147	1.94	48	0.63	48	0.63	113	1.49

* Includes all live and dead cases with these defects on the Registry at the end of 1964 excepting cases born in 1964.

a function of the perinatal mortality factor associated with the defect. This is illustrated in the following three examples of the use of Registry data:

(1) THE REGISTRY AS A SINGLE SOURCE OF INFORMATION—CLEFT LIP AND PALATE FINDINGS

The Registry by itself is a relatively good source of information for gross malformations with a relatively low stillbirth rate. The incidence rate of a malformation of this type is often expressed as the number of affected live-born children per unit number of live births. The cleft lip and palate complex is a good example.

The estimated incidence rate of cleft lip and/or palate in British Columbia based solely on Registry data is in good agreement with estimates by various authors published recently by Milham (1963) and shown in Table III. It is interesting to note that Milham's own rate of 1.80 per 1,000 births, obtained by combining information from birth and hospital records in Upper New York State, was the same as the rate per 1,000 live births for British Columbia. This rate was similar to rates obtained in Denmark (Fogh-Andersen, 1961) and Finland (Gylling and Soivio, 1962) which Milham (1963) claimed were fortunate from the stand-point of complete reporting.

On the whole, the estimates which were based solely on birth records in Table III are lower than

those derived from other sources. The exception to this is the estimate by Henderson (1940) based on birth record information in Hawaii. This, however, is not surprising, since the incidence of this condition is high among the Mongolian peoples, according to Miller (1964) and Neel (1958), and individuals of Japanese and Chinese ancestry constitute a large proportion of the population of Hawaii.

Greene (1963), using data from various sources, found that the combination of cleft palate and cleft lip occurred more frequently than either single component. He also found that cleft lip only and the combination were more common in the male while the reverse was true in the case of cleft palate only. Table IV (overleaf), showing sex-specific incidence patterns of the different types of buccal cavity deformity based on Registry data, lends partial support to these findings. The rates are underestimated for reasons explained in the Table footnote, but the relative differences are for the most part evident.

(2) REGISTRY DATA SUPPLEMENTED BY DEATH RECORDS—CONGENITAL HEART DISEASE FINDINGS

The type of malformation which most typically falls into this category is one in which the majority of cases are discovered after the time of birth but, because of the occult nature of the defect and a high

TABLE III
ESTIMATED INCIDENCE RATES OF CLEFT LIP AND/OR CLEFT PALATE

Years Surveyed	Investigator	Date	Source of Cases	Number of Cases	Number of Births*	Rate per 1,000
1953-57	Fogh-Andersen	1961	Surgical records of Diakonissestiftelsens Hospital, Copenhagen, Denmark	644	393,457 LB	1.64
1939-54	Gylling and Soivio	1962	Birth records, School of Midwifery, Helsinki, Finland	80	43,461 LB	1.84
1940	Henderson	1940	Birth records, Hawaii	35	18,024 LB	1.94
1951-55	Ivy	1957	Birth records, State of Pennsylvania	1,269	1,201,551 LB	1.06
1955	Loretz, Westmoreland, and Richards	1961	Birth records, State of California	368	313,164 LB	1.18
1940-50	McMahon and McKeown	1953	Registers of stillbirths and infant deaths; hospital records in Birmingham, England	285	218,693 TB	1.30
1944-57	Pleydell	1960	Register of hospital births, domiciliary births, infant deaths, and school health department registers, Northamptonshire, England	101	62,224 TB	1.62
1945-57	Rank and Thompson	1960	Plastic surgery clinic cases, doctors' reports, death records, hospital records, Tasmania	160	96,510 LB	1.66
1953	Wallace, Hoenig, and Rich	1956	Birth records, New York City	135	161,499 LB	0.84
1950-60	Milham	1963	Birth records and hospital records, Upper New York State	143	79,536 TB	1.80
1952-57	Present Study		Registry for Handicapped Children and Adults, British Columbia—1964 Live and Dead Caseload	367	203,642 LB	1.80

* LB—live births; TB—total births.

TABLE IV
ESTIMATED SEX-SPECIFIC "INCIDENCE" RATES OF CLEFT LIP AND PALATE, BY TYPE OF DEFECT
Based on the 1964 Live and Dead Registry Caseload

Type of Defect	Number of Cases Born in B. C. in the Years 1952-63			Rate per 1,000 Live Births occurring in B. C., 1952-63		
	Male	Female	Total	Male	Female	Total
Cleft palate only	109	114	223	0.49	0.53	0.51
Cleft lip only	112	53	165	0.50	0.25	0.38
Cleft palate with Cleft lip	220	125	345	0.98	0.58	0.79
Total Cleft palate and/or Cleft lip	441	292	733	1.97	1.37	1.68*
Number of Live Births	223,806	213,697	437,503			

* Slightly lower than the rate of 1.80 for 1952 to 1957 shown in Table III, because of lower rates in the more recent years resulting from a shorter casefinding period. The longer span of time represented in the above Table was necessary in order to obtain sufficiently large numbers in the sub-categories.

early mortality rate associated with it, many cases are discovered only at necropsy.

Access to death records is therefore important for estimating the incidence of this type of malformation. Congenital heart disease is a good example. Osterud, Menashe, and Martin (1965) in Oregon recently found $2\frac{1}{2}$ times as many cases with cardiovascular malformations on death certificates as on birth records relating to the same cohort of births. They also found that only 25 per cent. of the number of cases reported on death certificates had previously been reported on birth records. The relative importance of death records when ascertainment is based exclusively on vital records is seen in the Oregon cohort data in Table V, which shows 65 per cent. of all cases to be derived from this source. In British Columbia death records do not play so important a role, since many cases which are missed at birth are registered before they die. However, there is always a residue of cases which remain undetected until death. Cases derived from death records constitute more than 20 per cent. of the total number of British

Columbia cases in Table V. It is evident that the much higher overall rate per 1,000 live births in the British Columbia cohort is a result of augmentation by Registry data.

(3) REGISTRY DATA SUPPLEMENTED BY STILLBIRTH RECORDS—SPINA BIFIDA AND HYDROCEPHALUS FINDINGS

Another group of defects for which the Registry data must be supplemented is that of malformations of the central nervous system, such as spina bifida and hydrocephalus, in which stillbirths constitute a large proportion of the total number of affected births.

Gittelsohn and Milham (1962), for example, found that 15 per cent. of the spina bifida cases and almost 60 per cent. of the hydrocephalus cases reported in Upper New York State were stillbirths. Because of the large proportion of stillbirths, incidence rates of malformations such as these are usually expressed as rates per unit number of total live and stillbirths. In order to compare the Upper New York State rates based on total births with the corresponding rates in

TABLE V
DIFFERENT SOURCES IN CONGENITAL HEART DISEASE CASEFINDING
An Illustration of Relative Importance Using British Columbia and Oregon Data

Source of Data	British Columbia (1955-57 Cohort of 109,123 live births)			State of Oregon (1957-61 Cohort of 186,579 live births)*		
	No. of C.H.D. cases	Per cent.	Rate per 1,000 live births	No. of C.H.D. cases	Per cent.	Rate per 1,000 live births
Live birth records	42	7.5	0.38	144	34.7	0.77
British Columbia Registry cases not reported at birth (as at end of 1964)	392	70.1	3.59	—	—	—
Death records (reported for the first time at death)	125	22.4	1.15	271*	65.3	1.45
Total discovered cases in cohort (to end of casefinding period)	559	100	5.12	415	100	2.22

* The data included only cases born and dying in the period 1957 to 1961.

British Columbia, it was necessary to obtain from the Division of Vital Statistics the number of stillbirths with spina bifida and with hydrocephalus born in the period 1952-57 and to add these to the totals shown in Table I. The comparison is shown in Table VIA.

While the overall rates per 1,000 births of 1.14 (British Columbia) and 1.18 (Upper New York State) were similar in the two series in the case of spina bifida, the hydrocephalus rate was 33 per cent. higher in the British Columbia data (1.12 as compared with 0.84). Gittelsohn and Milham (1962), who used birth and death records, noted that hydrocephalus presents a special problem because it may be post-natal in origin. They also suggested that, since many cases developing after birth would have poor viability, they were subject to ascertainment through death records. However, the data indicate that later registration of children missed at birth probably accounted for the higher hydrocephalus rate in British Columbia.

This is brought into sharper focus if live-born and stillborn hydrocephalus rates per 1,000 total births are considered separately. The live-born rate of 0.79 in the British Columbia series was about double the

Upper New York rate of 0.35. On the other hand the stillborn rate in the British Columbia series was only 0.33 as compared with 0.49 in the Upper New York series. This was probably due in part to the fact that the Upper New York data included foetuses of 20 weeks or more gestation, while the limit was 28 weeks in the British Columbia data. (This was so prior to 1963 when foetuses of 20-27 weeks gestation were included for the first time.)

The apparent understatement of live-born hydrocephalus cases in the Upper New York series probably accounted for the wide disparity between the two series with respect to the percentage distribution of live and stillborn cases in Table VIA. This is in marked contrast to the almost identical distributions of spina bifida cases in the two series. However, it is reasonable to assume that inclusion of stillbirths of 20-27 weeks gestation in the British Columbia data would have increased the percentage of stillbirths among spina bifida cases.

As in other studies the female incidence rate for spina bifida was significantly higher than the male rate in both the British Columbia and the Upper New York studies, as shown in Table VIB. The

TABLE VIA
RECENT RECORDS OF SPINA BIFIDA AND HYDROCEPHALUS
Among Live and Stillbirths in Upper New York State and British Columbia

Type of Birth		Upper New York State (1945-59 Cohort of 2,552,910 births, including stillbirths of 20 weeks or more gestation)				British Columbia (1952-57 Cohort of 205,981 births, including 2,339 stillbirths of 28 weeks or more gestation)			
		Spina bifida		Hydrocephalus		Spina bifida		Hydrocephalus	
		No. of Cases*	Per cent.	No. of Cases*	Per cent.	No. of Cases†	Per cent.	No. of Cases†	Per cent.
Live		2,558	84.7	886	41.5	199	85.0	163	70.9
Still		463	15.3	1,251	58.5	35	15.0	67	29.1
Live and Still		3,021	100	2,137	100	234	100	230	100
Rate per 1,000 Total Live and Stillbirths	Live born cases	1.00		0.35		0.97		0.79	
	Stillborn cases	0.18		0.49		0.17		0.33	
	Live and Stillborn Cases	1.18		0.84		1.14		1.12	

* From birth records and death records of children under 5 years old.

† From live and dead Registry caseload at the end of 1964 and stillbirth records.

TABLE VIB
ESTIMATES OF SEX-SPECIFIC INCIDENCE RATES OF SPINA BIFIDA AND HYDROCEPHALUS
Upper New York State and British Columbia Data

Malformation	Upper New York State				British Columbia			
	Number		Rate per 1,000 births		Number		Rate per 1,000 Births	
	Male	Female	Male	Female	Male	Female	Male	Female
Spina bifida	1,304	1,709	0.99	1.38	104	130	0.99	1.29
Hydrocephalus	1,166	965	0.89	0.78	127	103	1.20	1.02
Total Number of Births	1,311.0*	1,242.0*			105,420	100,561		

* Approximate, in thousands.

female to male ratio was 1·39:1 in the Upper New York data and 1·30:1 in the British Columbia data. In the case of hydrocephalus, the male rate was higher in both series, although the difference was not statistically significant in the British Columbia data. However, when Gittelsohn and Milham (1962) separated the Upper New York population into white and non-white components, the latter comprising only 4 per cent. of the total number of births, the male rate for hydrocephalus was found to be higher in the white race but lower among non-whites. The higher female incidence of spina bifida, on the other hand, held true for both races.

FURTHER AUGMENTATION OF REGISTRY DATA BY VITAL RECORDS

As a result of the growing importance of the Registry as an information centre, a new recording system within the Registry organization was introduced in 1965 further to facilitate access to vital records and to ensure more comprehensive recording. By special arrangement with the central office of the Division of Vital Statistics, the Registry receives copies of live and stillborn records and death records relating to children under 7 years of age in which a congenital defect is mentioned. Information thus obtained is filed under a separate system after a careful check for duplication with the regular Registry caseload. The classification of defects and the organization of the data for research purposes are carefully supervised by two medical consultants and a genetic consultant to the Registry.

As a result of the broader basis of information which has thus been made possible, a more comprehensive programme of studying congenital defects has been undertaken. This involves the use of an expanded coding system, including aetiological and genetic sections, and will be the subject of a future communication.

SUMMARY

The use of a central registry for the handicapped in British Columbia for estimating the incidence of congenital defects is described. Although the Registry is a voluntary one, the large number of cases accumulated since its inception in 1952 has made it an important centre of information.

The main advantages of the Registry are:

- (1) Access to live birth records as a more or less direct source of registration of defects through its close working relationship with the Division of Vital Statistics.
- (2) A vast network of reporting agencies throughout British Columbia through which many

cases missed at birth are subsequently registered.

The augmentation of the former source of information by the latter is described with examples, congenital heart disease furnishing the most striking example.

The relative importance of the Registry and of different kinds of vital records for ascertainment in relation to different types of defect is discussed.

For gross malformations with relatively low perinatal mortality rates, the Registry has been used as an exclusive source of information. Estimates of the incidence of the cleft lip and palate complex based exclusively on Registry data are in good agreement with other published estimates.

For congenital defects of a more occult nature and with high post-natal mortality rates, Registry data must be supplemented by death records. Estimation of the incidence of congenital heart disease based on Registry data thus supplemented is more reliable than estimation based exclusively on birth and death records.

For severe defects with a high stillbirth rate, Registry data must be supplemented by stillbirth records. The use of the Registry in this manner for ascertaining the incidence of spina bifida and hydrocephalus is described.

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