Completeness of Birth Certificate Reporting for Mongolism (Down's Syndrome)

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UNDER-REPORTING of malformations on birth certificates is a recognized problem. The causes underlying such omissions are multiple and include failure or delay in diagnosis and lack of accuracy and completeness in filling out the certificate. Correspondence between the clinical record and the birth certificate is often poor, suggesting that underreporting of conditions of infants may result partly from the procedure of registration rather than from physicians' errors.

In several studies the accuracy and completeness with which the information recorded in the hospital record appears on the birth certificate have been compared (*1*, *2*). These investigations were limited to births occurring during relatively short time periods. In some the number of events has been large enough to include maternal complications as well as conditions of infants, while another approach has restricted the inquiry to specified malformations generally readily diagnosed at birth (*3*).

The primary documents used in this study are the birth certificates of institutionalized patients with a specific defect, Down's syndrome (mongolism). The aim was to determine the completeness of reporting of the condition at birth, with analysis to demonstrate possible biases in reporting caused by maternal age, population size of place of birth, birth weight, presence of other defects, the degree of malformation (the number of signs of Down's syndrome present among a subsample of patients studied), and time trends in reporting.

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Procedure

All patients with the diagnosis of Down's syndrome (mongolism) resident at Sonoma State Hospital, Eldridge, Calif., on January 1, 1962, were listed. Only those born in California were studied further. Copies of birth certificates were obtained from the office of vital statistics, California State Department of Public Health. The following information was taken from the birth certificate: maternal age, birth weight, mention of congenital malformation and maternal complications, and place and date of birth.

Cases of those born before 1933 were not analyzed because supplemental reporting of congenital malformations was not then required on the certificates. Hospital procedures preclude admission of most patients with this defect who are under 6 years; few patients born after 1956, therefore, were available for study. Patients born between 1950 and 1956 were analyzed in greater detail because they formed the largest group, and the nearly 50 percent completeness of reporting on their birth certificates provided adequate subgroups for comparisons.

Results

Table 1 presents a distribution of patients by place of birth (California or elsewhere), sex, and availability of the birth certificate. The predominance of males follows the pattern

among institutionalized defectives generally. A birth certificate was found for 95 percent of the California-born.

The completeness of reporting is shown by sex and time in table 2. No sex differences are present. However, the increased reporting in the 1950 to 1956 period and since 1956, compared with reporting in 1933–39, is noteworthy.

The following analyses were performed only for the 134 patients born between 1950 and 1956.

Comparison of those patients with Down's syndrome noted on the birth certificate with those without such mention for maternal ages at birth showed no significant difference (t= 1.818). Failure to record the mother's age occurred only twice in the 134 documents examined.

Comparison of birth-certificate reporting by population of birth place and sex is shown in table 3. For cities of more than 250,000 in 1950, the reporting and nonreporting of Down's syndrome on the birth certificate was equally complete, while those with between 100,000 and 249,999 inhabitants had a somewhat greater ratio (3 to 1) of reported to nonreported cases. The number of cases from areas with less than 100,000 population is smaller; however, the completeness of reporting generally is not greatly different from that of the largest communities, showing a ratio of 24 certificates with a diagnosis to 34 without.

A total of 43 of the 134 patients born between 1950 and 1956, selected at random, were studied for certain clinical features. Eighteen patients were drawn from the group with a diagnosis of Down's syndrome on the birth certificate and the remaining 25 from those without mention of the defect recorded at birth (table 4). The age range at the time of examination for these 43 patients was between 9 and 12 years. Ten cardinal signs of Down's syndrome (*4*) were chosen as representing the extent of physical characteristics which, taken in appropriate combination, are diagnostic of the syndrome. Certain of these physical abnormalities are age-dependent; irregular dentition, fissured tongue, and hyperflexibility are either not present or difficult to evaluate at birth, while epicanthus disappears with age. The average number of signs per patient, both sexes combined, was identical for the birth-certificate reported and nonreported groups, and for each sign, the similarity between the two groups was close.

No difference in birth-certificate reporting of Down's syndrome by birth weight was noted. Where the division was made at 5.5 pounds, 15 patients had birth weights under 5.5 pounds, and of these 7 were diagnosed on the birth certificate and 8 were not.

Table 1. Patients with Down's syndrome in Sonoma State Hospital (Calif.) on January 1, 1962, by birth State, sex, and birth certificate status

Category	Male	Female	Total
Born in California	204	163	367
Birth certificate	198	149	347
No birth certificate	6	14	20
Born elsewhere	54	22	76
Total	258	185	443

Table 2. Reporting of Down's syndrome on birth certificates, by time and sex

Year of birth	Male				Female				Both sexes			
	Reported		Not reported	Total	Reported		Not reported	Total	Reported		Not reported	Total
	Number	Percent			Number	Percent			Number	Percent		
1932 and before				20				24				44
1933–39	0	0.0	19	19	0	0.0	15	15	0	0.0	34	34
1940–49	9	12.7	62	71	7	14.0	43	50	16	13.2	105	121
1950–56	36	46.2	42	78	28	50.0	28	56	64	47.8	70	134
1956–61	8	80.0	2	10	1	25.0	3	4	9	64.3	5	14
1933–61	53	29.8	125	178	36	28.8	89	125	89	29.4	214	303

Incidental information on complications of pregnancy and labor, other congenital malformations, birth injury, and operations for delivery, by sex and reporting of Down's syndrome for the group born between 1950–56 is given in table 5. Overall, no major differences between this incidental information and the extent of reporting for the major abnormality are apparent. The completeness of data other than that concerning cesarean section is questionable (*2*).

Several sibships with more than one affected child are represented in the sample. Two male siblings with an admitting diagnosis of Down's syndrome were both identified correctly on the birth certificate, one born in 1954 and the second in 1959. In another sibship the first born affected (1951) was so indicated on the birth certificate whereas a brother, born in 1953, was not reported. In a third family, the birth certificate of a female born in 1953 did not have mention of Down's syndrome whereas an older

Table 3. Reporting of Down's syndrome on birth certificates, by population of birth place, sex, and completeness of reporting, sample born 1950–56

Sex	Population size of birth place in 1950							
	Over 250, 000	100, 000–249, 999	50, 000–99, 999	25, 000–49, 999	10, 000–24, 999	2, 000–9, 999	Other	Total
Males	29	19	2	13	8	3	4	78
Reported	12	13	1	6	2	0	2	36
Not reported	17	6	1	7	6	3	2	42
Females	21	7	4	7	9	2	6	56
Reported	9	6	3	3	4	0	3	28
Not reported	12	1	1	4	5	2	3	28
Both sexes	50	26	6	20	17	5	10	134
Reported	21	19	4	9	6	0	5	64
Not reported	29	7	2	11	11	5	5	70

Table 4. Distribution of physical signs and other clinical characteristics of 43 patients with Down's syndrome, born 1950–56, by sex and completeness of reporting on birth certificates

Clinical characteristic	Male		Female		Both sexes			
	Reported	Not reported	Reported	Not reported	Reported		Not reported	
					Number	Frequency	Number	Frequency
Number of patients	10	14	8	11	18	-------	25	-------
Average age in 1962 (years)	9. 9	10. 2	10. 2	11. 0	10. 0	-------	10. 6	-------
Average age at admission (years)	2. 1	4. 4	2. 6	5. 6	2. 3	-------	4. 9	-------
Average IQ	20	26	20	29	20	-------	27	-------
Average number of signs per patient [1]	8. 0	8. 5	9. 0	8. 1	8. 4	-------	8. 4	-------
Epicanthus	6	12	7	9	13	0. 72	21	0. 84
Oblique palpebral fissures	7	8	7	10	14	. 78	18	. 72
Protruding tongue	4	6	1	4	5	. 28	10	. 40
Flat nose bridge	9	11	8	10	17	. 94	21	. 84
Simian line	5	10	6	6	11	. 61	16	. 64
Short, incurved fifth finger	9	12	8	10	17	. 94	22	. 88
Hyperflexibility	10	14	8	9	18	1. 00	23	. 92
Irregular dentition	7	11	7	7	14	. 78	18	. 72
Brushfield spots	7	10	4	7	11	. 61	17	. 68
Fissured tongue	8	12	6	8	14	. 78	20	. 80

[1] Ten cardinal signs after Oster, reference 4.

Table 5. Supplemental information on birth certificates of 134 patients with Down's syndrome, born 1950–56, by sex and completeness of reporting

Supplemental information	Male		Female		Both sexes	
	Reported (N=36)	Not reported (N=42)	Reported (N=28)	Not reported (N=28)	Reported (N=64)	Not reported (N=70)
Complications of pregnancy and labor	2	6	4	5	6	11
Breech	0	2	1	2	1	4
Cephalic-pelvic disproportion	[1] 1	[1] 1	[2] 1	0	2	1
Myomas	0	[1] 1	[2] 1	0	1	1
Retained placenta	0	0	0	1	0	1
Premature labor	0	1	1	1	1	2
Eclampsia	0	0	0	1	0	1
Polyhydramnious	1	0	0	0	1	0
Bleeding, third trimester	0	[3] 1	0	0	0	1
Other congenital malformations	4	2	2	0	6	2
Cardiac defect	0	1	1	0	1	1
Erythroblastosis fetalis	1	0	1	0	2	0
Accessory signs of Down's syndrome	1	0	0	0	1	0
Clubfoot, type not specified	1	0	0	0	1	0
Talipes varus	1	0	0	0	1	0
"Flap ears"	0	[3] 1	0	0	0	1
Birth injuries	0	0	0	0	0	0
Cesarean section	[1] 3	[1] 5	[2] 2	1	5	6
Total, all supplemental information	9	13	8	6	17	19

[1] Cephalic-pelvic disproportion and cesarean section reported on same certificate.
[2] Cephalic-pelvic disproportion, myomas, and cesarean section reported on same certificate.
[3] Bleeding during third trimester and "flap ears" reported on same certificate.

male sibling, born in 1951, was later admitted to another California institution with a diagnosis of Down's syndrome. The birth certificate for this child was not available.

Discussion

Study of a large group of institutionalized patients with Down's syndrome suggests that under-reporting of this condition on the birth certificate is appreciable. However, improvement has been shown in the accuracy of this information over time for the study group. Between the periods 1933–39 and 1950–56, the completeness of reporting increased from 0 to about 50 percent.

The complete absence of reporting in the group born between 1933 and 1939 may partly reflect a lag following the introduction of supplemental reporting in 1933. Further, the shift in the impact on the population of different diseases between 1933 and 1956 has resulted, in recent years, in an increased awareness by the medical community of the importance of chronic conditions diagnosable at birth.

Because the study population consisted of surviving patients with Down's syndrome resident in a State hospital for the mentally retarded, the results reported cannot be compared exactly with those published studies in which ascertainment was either from hospital birth records or within a series of all births in a large population group. Babbot and Ingalls (*3*) have recently reported that out of 26 cases of Down's syndrome noted in the hospital birth record 15, or 58 percent, were also reported on the birth certificate. The sample was drawn during 1955–60 from a large hospital in a major city and from a general hospital in a less densely populated area. No difference was noted in the completeness of reporting for Down's syndrome between these two places. The 58 percent reporting approximates the value of 48 percent for the subgroup born between 1950 and 1956 and discussed here.

In a study of the possible association between congenital malformations and low radiation levels, Gentry and associates (*5*) surveyed all birth certificates and all deaths of children un-

der 5 years of age from 1948 to 1955 in New York State exclusive of New York City. The reported rate for Down's syndrome was 0.3 per 1,000 live births. Comparison with a projected rate of 1.45 per 1,000 live births (*6*) suggests that only approximately 20 percent of cases were recorded on the birth certificates. This result may indicate that the 50 percent completeness in reporting noted for the sample born during 1950–56 and reported here is too high and that it represents bias in some direction associated with institutionalization and not revealed in the analysis.

From a study of major congenital malformations in Japanese infants conducted as part of the genetics program of the Atomic Bomb Casualty Commission, Neel (*7*) has reported an incidence of Down's syndrome diagnosed at birth or on re-examination at 8 to 10 months of age of 0.87 per 1,000 live births. Compared to a pooled estimate of 1.45 per 1,000 live births, the value Neel reported is 60 percent of expectation. Further, of the 14 cases in the sample undergoing the 8 to 10 months' re-examination, 12 were diagnosed at followup.

The difficulty of diagnosing Down's syndrome at birth has been discussed by Carter and MacCarthy (*8*). Case identification is complicated and proper diagnosis would depend upon the experience of the examiner, the degree of phenotypic expression, and other factors. However, intensive study of infants such as the Japanese investigation, focused specifically on congenital malformations, are unlikely to be incomplete. When failure of diagnosis at birth is a major source of under-reporting, as in Down's syndrome, re-examination later could give a false low rate owing to a high mortality in early infancy of those affected (*9*). Inquiry into many deaths among the study group of Japanese infants, without identification of missed cases, appears to eliminate this bias. A possibility that incidence rates of Down's syndrome do vary in time and place thus remains.

The recently discovered chromosomal abnormality of Down's syndrome has reinforced the importance of studies into the etiology and pathogenesis of this malformation (*10*). The presence of the extra chromosome provides a study opportunity for a variety of genetic investigations. For the epidemiologist, however, description of the distribution of the disease in time and space, by host characteristics and by association with other variables, demands accurate and nearly complete ascertainment of cases in population samples of large size. A few published studies (*6*, *11*) suggest that the distribution of cases is nonrandom in association with several variables. In a recent review Cohen and co-workers (*12*) presented a range of incidence rates wide enough to invite further epidemiologic studies. In designing such studies, the problem of under-reporting on birth certificates necessitates the use of many case-finding techniques. Results from this investigation and the others discussed here suggest that reporting on birth certificates is from 20 to 50 percent complete. Further, lack of differentiation between birth-certificate reported and nonreported cases in analyses of some familial, clinical, and demographic characteristics suggests no improvements in the currently used forms and methods or in identifying missed cases by positive responses other than mention of Down's syndrome as such on the congenital malformation portion of the document.

Summary

A survey of birth certificates of institutionalized patients with a clinical diagnosis of Down's syndrome has indicated that under-reporting of this condition at birth is appreciable. However, the completeness of reporting for the group studied varied in time, being 0 percent for those patients born in the 1933–39 period and almost 50 percent for the group born in the 1950–56 period. Analysis of cases of those born between 1950 and 1956, the largest subsample, by maternal age at parturition, place of birth, birth weight, clinical signs of Down's syndrome on current examination, and, where recorded, complications of labor and delivery and other congenital malformations did not reveal any differences between the groups with and without mention of a diagnosis on the birth certificate.

No other study has been reported in which diagnosed surviving patients with Down's syndrome were matched to the birth certificate. Under-reporting of this malformation, however, is apparent in several studies utilizing other methods of case identification. Between 20 and

50 percent completeness of reporting has been noted. Variations in the incidence of Down's syndrome in time and space could be obscured by incomplete casefinding, an important complication in the design and analysis of epidemiologic studies of this malformation.

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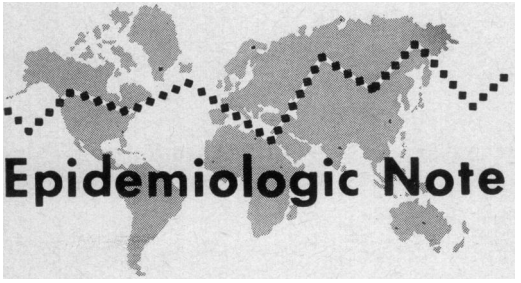

New Immigrant Mosquito in Hawaii

Inadvertent introduction of a mosquito species into Hawaii, the first since the turn of the century and the days of the sailing vessel, was detected on January 2, 1962, when a single specimen of *Aedes vexans nocturnus* (Theobald) was found in a light trap catch at the Public Health Service Quarantine Station in Honolulu. Subsequently, a heavy incidence of adults and larvae was discovered on the Ewa side of Pearl Harbor. Because the species is a potential vector, civilian and military agencies immediately moved to determine the extent of infestation and to prevent the rapid spread of the species to other parts of Oahu and neighboring islands.

Later surveys recorded the spread of the species throughout most of Oahu from Waimanalo to Kahuku, Waialua, and Waianae. It is now well established also on the neighboring island of Kauai.

The species, a potential vector of Japanese B encephalitis, has been intercepted a number of times on aircraft through quarantine inspection. The source of the introduction may be Guam, Samoa, Fiji, Philippines, or the Marshall Islands, since all have ports of departure for aircraft and ships coming to Hawaii.

Mosquitoes were unknown in Hawaii until 1826. Today, *Aedes aegypti, Aedes albopictus, Aedes vexans nocturnus, Culex quinquefasciatus,* and two purposely introduced *Toxorhynchites* species are present.—C. R. JOYCE, *scientist director, Public Health Service Quarantine Station, Honolulu,* and P. Y. NAKAGAWA, *chief, mosquito control, vector control branch, Hawaii State Department of Health.*