*An incidence and prevalence study of a wide range of long-term conditions of children was carried out in Erie County, New York. Its purpose was to provide a basis for expanding and improving an existing rehabilitation program, and to obtain basic epidemiologic data. Based on hospital records, office records of medical specialists, school records, and vital records of official agencies, this intensive study will provide valuable information. Various aspects, including advantages and deficiencies, are discussed, and possible applications are touched on, for example the incidence of cystic fibrosis.*

# THE ERIE COUNTY SURVEY OF LONG-TERM CHILDHOOD ILLNESS: I. METHODOLOGY

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ALTHOUGH recent research has greatly advanced the management of many long-term childhood conditions, the need for organized programs of care for affected children has become increasingly apparent to physicians, health agencies, and the general public. They have urged government aid for families heavily burdened by the expenses involved. Assistance is also needed to insure payment for care to underfinanced medical facilities and to encourage the development of additional facilities in those areas where they do not yet exist. In 1961, New York State proposed an approach to a solution through an extension of its children's medical rehabilitation program. Since a review of literature yielded inadequate information for the expansion, a study was planned to supply data relative to occurrence and distribution of the childhood conditions. Such a study would have much broader epidemiological importance than the immediate program needs within New York State.

In June, 1962, the New York State Department of Health initiated a three-year incidence and prevalence study of some 70 long-term childhood diseases not then covered by its medical rehabilitation program. The project was conducted under the auspices of the Department of Health of the county of Erie, which employed the study personnel and administered the funds which the New York State Health Department provided for the project. Although primarily concerned with asthma, muscular dystrophy, cystic fibrosis, nephrosis, diabetes, and blood dyscrasias, the project also included celiac disease, arthritis, and other diseases for which aid was not yet available. Preliminary findings from the survey were used in planning the expanded medical rehabilita-

tion program[1] for the care of children with long-term conditions and illnesses.

The study attempted to answer the following questions:

1. What is the incidence and prevalence of illness among children whose conditions are classified as "long-term" and sufficiently serious to necessitate medical care?
2. How are the incidence and prevalence of long-term illness in children related to race, economic level, and other social and economic variables?
3. What further need is there for medical and ancillary facilities and services to treat and rehabilitate the chronically ill child?
4. What is the impact of long-term illness on the child and his family, and what factors, preexisting and concomitant, influence these effects?

Erie County, New York, was designated as the sample area. Its population of over 1,000,000 was considered adequate for case finding even for those conditions whose occurrence is relatively rare. Both urban and rural residents made up a comparatively stable population in many ways representative of the state. Medical care facilities in Buffalo, the major city of the county, were considerable and well developed, providing treatment for all conditions under study; and very little medical care for Erie County residents was obtained outside the county.

## Study Plan

The study plan called for the identification of every child in Erie County diagnosed as having one of the study diseases before the age of 16 and the determination of pertinent epidemiological data relative to the patient and his family. A survey of medical records covering a time period of 16 years was chosen so that the researchers could ascertain any hospitalizations occurring even in infancy for all the children under study. Because of the lag in classifying records in the hospitals to be surveyed, the years 1946 through 1961 were selected.

The study included the following diseases and conditions. The assignment to specific categories is necessarily arbitrary in some instances:

Allergic Disorders
- Asthma
- Eczema

Endocrine Disorders
- Cretinism
- Hypoglycemia, leucine induced
- Diabetes mellitus
- Hyperparathyroidism
- Hypoparathyroidism, chronic idiopathic
- Pseudohypoparathyroidism
- Diabetes insipidus
- Pituitary dysfunction
- Adrenal hyperplasia
- Adrenogenital syndrome

Respiratory System
- Bronchiectasis

Metabolic Diseases
- Albinism
- Celiac syndrome
- Eosinophilic granuloma
- Hand-Schuller-Christian syndrome
- Gaucher's disease
- Niemann-Pick disease
- Amaurotic familial idiocy (Tay-Sachs disease)
- Gargoylism
- Hemosiderosis
- Erythropoietic porphyria, congenital
- Phenylketonuria (PKU)
- Alkaptonuria
- Maple syrup urine disease
- Fructosuria
- Penosuria
- Sucrosuria
- Galactosemia
- Glycogen storage disease
- Glycogenosis
- Franconi-DeToni syndrome
- Cystic fibrosis of the pancreas
- Cystinuria
- Cystinosis
- Letterer-Siwe disease
- Tyrosinosis
- Hypophosphatasia
- Analbuminemia
- Hartnup disease
- Kernicterus, with non-hemolytic jaundice
- Marfan's disease
- Nonhemolytic jaundice (Gilbert's disease)

Diseases of the Blood and Blood Forming Organs and Circulatory System
- Lupus erythematosus
- Hageman trait

Spherocytosis
Anemia, hemolytic
Hypoplastic anemia, congenital microspherocytosis
Agammaglobulinemia
Afibrinogenemia
Hemophilia A, deficiency of anti-hemophilic globulin
Hemophilia C
Christmas disease
Thalassemia
Sickle cell anemia
Hemoglobins, abnormal
Purpura, anaphylactoid
Purpura, non-thrombocytopenic
Methemoglobinemia
Periarteritis nodosa
Pseudohemophilia

Central Nervous System
Encephalitis
Encephalomyelitis
Disseminated sclerosis
Wilson's disease (hepatolenticular degeneration)
Huntington's chorea
Friedreich's ataxia
Hereditary periodic paralysis

Digestive System
Ulcer, peptic
Colitis, ulcerative
Liver cirrhosis

Genito-Urinary System
Nephrotic syndrome
Nephritis
Renal rickets (resistant rickets)
Renal dwarfism
Renal infantilism
Albright's syndrome (renal tubular acidosis)

Bone and Organs of Movement
arthritis, rheumatoid
dermatomyositis
muscular dystrophy
muscular atrophy
amyotonia congenita
myotonia congenita

Because of the relative rarity of many of the conditions and the necessity of valid medical diagnoses in all cases, household survey methods, used in previous projects[2-4] with similar objectives, were not used as the basis for this study. Instead, the information was sought in four types of medical records: hospital charts, specialist physicians' office records, birth and death certificates, and selected school health records. Additional demographic and social data were obtained from home interviews of a sample of the study population.

During the hospital-survey phase of the study, project personnel worked in the medical record room of each hospital with cooperative assistance from its librarian. From the disease classification cards for 16 years, they selected and read the charts of all child patients whose discharge diagnoses indicated any of the study diseases. Appropriate information was transferred to forms, which were then returned to the study office. The Children's Hospital of Buffalo, a large university-affiliated teaching hospital, provided the greatest amount of data about the more serious conditions. The case-finding results were double checked there by comparison with admission books for the study years and the personal files which several hospital-staff specialists kept of patients with diseases in which they were particularly interested.

The degree of validity which could be assumed for this information was, however, limited by classification methods in the various medical-record libraries. In addition, many "possible" and "probable" diagnoses had to be included pending further investigation. An under-enumeration of disease incidence and prevalence resulted in three hospitals where disease classification cards had been destroyed for several years prior to 1956. Nevertheless, the results of the hospital survey were well worth the two years' work by trained study personnel.

Several of the conditions under study did not usually require hospitalization. Consequently, a private medical-office record survey was indicated to complete the incidence and prevalence estimates. This survey was limited to pediatricians, allergists, and other appropriate specialists because their treatment or consultative service would probably be re-

quired by almost all children with these diseases. After the Medical Society of the County of Erie had given its endorsement, a letter of explanation was sent to each specialist. The study director or his assistant then visited the office by appointment and explained in detail the methodology of the study. He requested permission to survey the physician's records and suggested several methods to effect the search. In most cases, the physician was aware of the amount of time and effort required and agreed to permit the study personnel access to his files. Unfortunately, since the records were always classified alphabetically rather than by diagnosis, registered nurses had to perform the time-consuming task of reading through the charts and selecting those with study diagnoses for the recording clerks. Also, in the few cases where physicians preferred to search their own files, it was impossible to insure that their contributions were complete. Although the idea of physicians allowing data to be abstracted from their private office records, even for statistical purposes, produced a great deal of discussion and some negative response, 40 of a possible 49 medical specialists cooperated with the study.

The collection process revealed two major problems: First, hospital and medical records, not primarily designed for research purposes, included many incomplete, illegible, and unsystematized charts. Second, diagnostic variability among physicians had to be accepted without standardization. Arbitrarily, discharge diagnoses were not questioned unless qualified by "possible," "probable," or some similar indication of an uncertain or disputed diagnosis. The director of child health of the Erie County Health Department,* a board-certified pediatrician, reviewed those charts and advised as to the diagnostic designation of the cases. Home-interview information and death certificates were used later to correct partially any error resulting from this procedure. Fortunately, the vast majority of diagnoses were reinforced by a consensus of several medical opinions and by family awareness of the condition over years of care. Some were also substantiated by autopsy findings.

Prevalence estimates necessitated knowledge of the survivorship of the study population. This information was secured with the cooperation of the Office of Biostatistics and Vital Records of the New York State Department of Health. Records of all deaths among children residing in Erie County from 1946 through 1961 were extracted by computer from the vital records of New York State. A print out based on causes of death and death certificate numbers permitted selection of those deaths which might, in any way, relate to a study disease. Examination of the corresponding death certificate provided complete information, which was returned to the project office. These names were matched to study cases, and the associated mortality was established.

Student health records for six public schools with special classes for handicapped children were searched as an additional source of study cases.

A basic table for each disease was used to show yearly incidence, associated mortality, and cumulative prevalence. As an example, the basic data for cystic fibrosis are presented in Table 1.

The study objectives required home interviews of a sample of the chronically ill population to determine the impact of each disease on the children and their families, to ascertain their needs, and to gather further data on pertinent epidemiological variables. The sample selected for home interview was drawn from the total population of 2,183

* We are indebted to Ursula Anderson, M.D., D.P.H., whose consultation contributed significantly to this study.

chronically ill children less than 16 years of age as of December 31, 1961. Study children who had passed their sixteenth birthday were excluded to limit the recall period to the most recent childhood experiences.

## Sampling Procedure

Two sampling procedures were used. The first involved a random selection of ten cases from each disease category, or all the cases from those groups containing less than ten, so that each disease would be represented in the home interview sample. This first selection resulted in 292 cases. Since a potential sample of at least 500 cases was desired, 251 additional cases were selected at random from the remaining groups, which were pooled without regard to disease category. An over-sampling of 43 cases was included to compensate for potential loss.

The home interview schedule required about one and one-half hours to complete. The information which it elicited included demographic, social, and economic variables; hospital, physician, and clinic utilization; history of the study illness; medications administered; special care and diets required; any family history of the study disease; parental reaction to the diagnosis; education experience of the chronically ill child; changes in social activities; and the effect of the study disease on the relationship of the parents.

Arrangements for the home interview were begun by writing to the attending physician in each selected case. The letter concluded by stating that a telephone call would be made in a few days to determine whether, for any reason, he did not wish the family to be contacted. If the physician did not object, a letter was then sent to each family in the interview sample describing the study and informing them that we wished to conduct a home visit. A few

**Table 1—Incidence, prevalence and associated mortality of cystic fibrosis of the pancreas diagnosed during the years 1946 through 1961 among Erie County residents under age sixteen**

| Year | Old Cases | New Cases | Deaths | 16th Birthday | Total Cases at end of Year |
|---|---|---|---|---|---|
| 1946 | — | 2 | — | — | 2 |
| 1947 | 2 | 3 | 1 | — | 4 |
| 1948 | 4 | 2 | — | — | 6 |
| 1949 | 6 | 6 | 1 | — | 11 |
| 1950 | 11 | 5 | 5 | — | 11 |
| 1951 | 11 | 3 | 1 | — | 13 |
| 1952 | 13 | 5 | 2 | — | 16 |
| 1953 | 16 | 5 | 2 | — | 19 |
| 1954 | 19 | 9 | 4 | 1 | 23 |
| 1955 | 23 | 10 | 6 | 1 | 26 |
| 1956 | 26 | 11 | 5 | — | 32 |
| 1957 | 32 | 8 | 5 | — | 35 |
| 1958 | 35 | 8 | 3 | — | 40 |
| 1959 | 40 | 18 | 6 | — | 52 |
| 1960 | 52 | 8 | 11 | 2 | 47 |
| 1961 | 47 | 5 | 6 | — | 46 |
| | — | — | — | — | — |
| Total | — | 108 | 58 | 4 | — |

days later, an appointment was arranged by telephone.

One hundred and sixty-five, or approximately 92 per cent, of the 180 physicians listed as physicians of record for the cases in the sample responded affirmatively. Three physicians were deceased, one had moved from the area, four were no longer associated with the cases, four could not be contacted, and three were negative to the study. Most of those who cooperated gave additional assistance by providing recent addresses and other information about the families in question.

The project was presented to the families as a research study of childhood illness, including children who had been hospitalized at least once. At no time was the medical diagnosis mentioned, nor was there any reference to specific illnesses. The interview questions were designed to elicit a complete medical experience history without emphasizing a specific diagnosis. Interviewers received extensive training to deal with the particular sensitivity of those involved in the subjects under discussion.

## Related Literature

Another aspect of the study involved the compilation of pertinent literary references relating to incidence and prevalence; basic epidemiologic studies; and social, psychological, and nursing articles dealing with the impact of each specific disease on the family. Abstract collations and literature indexes were checked under each disease category. Pertinent articles published during the last ten years were read and summarized, and the summaries were transferred to McBee Keysort Cards and coded. Over 500 articles have been processed into this system, which allows for efficient literature retrieval of article summaries as well as immediate identification of pertinent publications. It will accommodate a great increase in the number of publications which can be summarized and added.

The incidence of diseases, especially those of a genetic nature, has long been of major interest to clinicians and epidemiologists and takes on added importance as the statistical basis for planning service programs. Estimates are often quoted from one publication to another with no investigation of the sources of the original data. Cystic fibrosis is a typical case in point.

Recently, in both scientific literature[5] and the popular press,[6] the incidence of cystic fibrosis has been stated as approximately one in 1,000 live births. More dramatically, it is often stated that a baby with cystic fibrosis is born every two and one-half hours. A review of the literature to find the source of these figures revealed that the original study was published in 1946 by Andersen and Hodges.[7] Their estimate was based on a survey of all autopsied children less than 14 years of age who had died with cystic fibrosis in several New York City hospitals. They calculated the total number of deaths by applying the frequency obtained from autopsies (3 per cent) to the total number of deaths among children less than 14 years of age in New York State in 1939. Steinberg and Brown pointed out in 1960[8] that this calculation involved the possibly incorrect assumption that "patients who died with cystic fibrosis are a random sample of all patients coming to autopsy and that patients coming to autopsy are a random sample of all who died." Goodman and Reed (1952)[9] stated their assumption that hospital autopsies represent a biased sample of infant deaths since many accidental deaths would probably not appear in hospital autopsy reports. Since Andersen and Hodges then related their autopsy data to live births, estimating incidence to be 1.7 deaths per 1,000 live births in New York State, it must be

added that there was an error in their base population of unknown magnitude since the population base used to estimate the frequency of cystic fibrosis was all live births in New York State for the one year 1939. This base population of one year's births appears too small an estimate of the population at risk since deaths had been computed for children born during a 14-year period.

Other investigators have used differing methods to determine the incidence of cystic fibrosis with varying results.[10-15] Goodman and Reed, in 1952, concluded that the incidence of cystic fibrosis approximated one in 1,000 live births. Steinberg and Brown, in a 1960 report, examined death certificates for all white children born in a four-year period in the state of Ohio and checked against hospital records all deaths which indicated the possibility of cystic fibrosis. They then elicited names, birth dates, and life status of cases of cystic fibrosis from all Ohio pediatricians, all major hospitals in the state and the Cleveland Cystic Fibrosis Foundation. When 198 cystic fibrosis patients had been identified and incidence rates determined for each county, there was no significant difference in rates among the counties. Steinberg and Brown estimated the incidence of cystic fibrosis as one in 3,700 white live births.

A more recent estimate was made by Kramm, Crane, Sirken, and Brown[16] from the results of a cystic fibrosis pilot survey conducted in Massachusetts, New Hampshire, and Vermont. In 1962, after a mail survey of private physicians and out-patient departments, they reported their preliminary estimates of the incidence of diagnosed cases of cystic fibrosis in the pilot area as 42 per 100,000 live births or one case per 2,300 live births.

The Steinberg and Brown, and the Kramm, Crane, Sirken, and Brown estimates, both far more recent and each based on an apparently logical approach, have generally been ignored in preference for the more dramatic figure resulting from studies conducted 13-20 years ago which had obvious biases inherent in their methodology.

The results of the childhood disease study conducted in Erie County under favorable conditions should provide a more accurate answer to the intriguing question of incidence, at least among white children. The incidence rate is based on a definition of the population at risk which includes all white children under ten years of age residing in Erie County in a single year as determined by the average population for the years 1957-1961. The number of new cases of cystic fibrosis was determined from the average of the number of new cases diagnosed per year for the same five-year period. From these estimates, the yearly incidence has been calculated to be 3.7 cases per 100,000 children under ten years of age or one case per 27,027 white children under ten years of age. In order to compare with the estimates mentioned previously, incidence per live births was obtained in the following manner: The 16-year experience in case finding indicated that 82 per cent of the reported cases were diagnosed when less than five years of age. By taking five yearly cohorts of live births and following them for five years, it was found that the average number of new cases diagnosed per cohort resulted in an incidence figure of one case per 2,941 live births.

Projecting this 82 per cent estimate to 100 per cent results in an incidence for cystic fibrosis of one in 2,439 live births. This incidence rate is higher than the rate of 1:3,700 live births determined by Steinberg and Brown, whose methodology is most similar to ours. It might be hypothesized that our case-finding procedure conducted more intensively in a smaller area was less likely to miss cases than a survey which attempted to enumerate every

**Table 2—Incidence of cystic fibrosis among yearly cohorts of live births in Erie County for five-year periods**

| Cohort | Year | Live Births | 5-Year Observation: Year Ending | 5-Year Observation: No. of Cases |
|---|---|---|---|---|
| 1 | 1953 | 22,369 | 1957 | 7 |
| 2 | 1954 | 23,824 | 1958 | 9 |
| 3 | 1955 | 24,492 | 1959 | 10 |
| 4 | 1956 | 25,084 | 1960 | 12 |
| 5 | 1957 | 26,212 | 1961 | 4 |
| Total | | 121,981 | | 42 |
| Yearly Average | | 24,396 | | 8.4 |

Incidence Rate $\frac{8.4}{24,396} = 0.00034$ or 1 per 2,941 live births

case diagnosed in an entire state. The estimate of one in 2,300 live births resulting from the Cystic Fibrosis Pilot Survey in Three New England States is comparable with that of the Erie County Childhood Disease Study. Although the former, like Steinberg and Brown's, covered a large area, cases were obtained from a more comprehensive array of sources and compensated for the probability of missing cases by including over 6 per cent "probable" diagnoses.

The Erie County study has overcome to a considerable degree a deficiency of previous studies: their inability to determine the initial population represented by the affected group. A more complete presentation and explanation of the cystic fibrosis data obtained in this study will be presented in a subsequent publication.

## Summary

An incidence and prevalence study of a broad group of long-term childhood diseases and conditions was conducted in Erie County, New York, for the purpose of obtaining basic epidemiologic data as an end in itself and as a basis for expanding and improving an already existing rehabilitation program for children. Conducted over a period of three and one-half years, this project attempted to identify each case of long-term disease diagnosed during the 16 years from 1946 to 1961, among residents of Erie County under age 16. Hospital charts, medical specialists' office records, certain school records, and the vital records of the state of New York all contributed to the study data.

Home interviews were conducted among parents of a sample of the recorded cases to gather additional demographic data and to gain insight into the effect of each disease on the child and his family, as well as to define more clearly the associated problems of care and social and academic adjustment.

The incidence of cystic fibrosis of the pancreas has been presented here as an example of the case-finding technic, with the resulting incidence and prevalence rates based on a well-defined population. The data indicate that the incidence of cystic fibrosis in Erie County, New York, approximates one in 2,500 live births.

The deficiencies of this study center primarily on variations in diagnosis for certain conditions and a recognized incompleteness of case enumeration as determined from even an intensive medical record survey.

## REFERENCES


1. Schlesinger, E. R.; Bahlke, A. M.; and Cohen, A. R. Development of a Statewide Program for the Care of Children with Long-term Illness. A.J.P.H. 55:973–977 (July), 1965.
2. Commission on Chronic Illness. Chronic Illness in the United States: Chronic Illness in a Large City, The Baltimore Study. Vol. IV. Cambridge, Mass.: Harvard University Press, 1957.
3. Trussell, R. E., and Elinson, J. Chronic Illness in the United States: Chronic Illness in a Rural Area, The Hunterdon Study. Vol. III. Cambridge, Mass.: Harvard University Press, 1959.
4. Schiffer, C. G., and Hunt, E. P. Illness among Children. U. S. Department of Health, Education, and Welfare, Children's Bureau, Publ. No. 405, 1963.
5. di Sant'Agnese, P. A. Cystic Fibrosis of the Pancreas. Health News 41:4–11,19 (Sept.), 1964.
6. ————. In Metabolic Disorders: Living with Cystic Fibrosis. Time 86, 8:43, 1965.
7. Andersen, D. H., and Hodges R. G. Celiac Syndrome V. Genetics of Cystic Fibrosis of the Pancreas with a Consideration of the Etiology. Am. J. Dis. Child. 72:62–80, 1946.
8. Steinberg, A. G., and Brown, D. C. On the Incidence of Cystic Fibrosis of the Pancreas. Am. J. Human Genet. 12:416–424, 1960.
9. Goodman, H. O., and Reed, S. C. Heredity of Fibrosis of the Pancreas. Ibid. 4:59–71, 1952.
10. Low, C. U.; May, C. D.; and Reed, S. C. Fibrosis of the Pancreas in Infants and Children. Am. J. Dis. Child. 78:349–374, 1949.
11. di Sant'Agnese, P. A. Cystic Fibrosis of the Pancreas. Am. J. Med. 21:406–422, 1956.
12. di Sant'Agnese, P. A., and Lowe, C. U. Cystic Fibrosis of the Pancreas: Excerpts from a Round-table Discussion. Pediatrics 19:1136–1138, 1957.
13. Andersen, D. H. Cystic Fibrosis of the Pancreas. J. Chronic Dis. 7:58–90, 1958.
14. di Sant'Agnese, P. A., and Andersen, D. H. Cystic Fibrosis of the Pancreas in Young Adults. Ann. Int. Med. 50:1321–1330, 1959.
15. Selander, P. The Frequency of Cystic Fibrosis of the Pancreas in Sweden. Acta paediat. 51:65–67, 1962.
16. Kramm, E. R.; Crane, M. M.; Sirken, M. G.; and Brown, M. L. A Cystic Fibrosis Pilot Survey in Three New England States. A.J.P.H. 52:2041–2057, 1962.


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This paper was submitted for publication in November, 1965.