Appendix 6.4

Data Source Described in Detail – Genetic Services

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Source or Site

- Regional/state genetics networks
- Hospital-based genetics clinics
- University-based genetics clinics
- Provider-based genetics clinics

Geneticists and dysmorphologists are skilled at evaluating a constellation of findings, providing differential diagnoses, and determining the definite medical condition. They use diagnostic procedures such as chromosomal analysis and genetic testing, as well as drawing from their personal experiences and extensive literature in evaluating a patient.

The information from this data source is of high quality.

Legal or Professional Mandates

- *Legal.* State statutes for hospital-based clinics. These are operated in accordance with hospital licensing and accreditation.
- *Legal contract.* Specified in individual contracts or collaborative agreements.
- *Professional.* Certification and professional credential as required.

Mission or Objective

Genetic diagnostic and counseling services, therapeutic management of genetic diseases.

Scope or Breadth

Clinics may include prenatal, pediatric, and/or general population. Some may be specialized by disease category (e.g., Down syndrome, cystic fibrosis). Some providers include diagnostic and research laboratories, clinical research centers, and off-site clinics.

Operational Structure

Genetics clinics may be set up as a referral site (i.e., to provide a diagnosis back to the referring physician), for services (i.e., for ongoing treatment and consultation), or for research or study (i.e., database).

Type of Information Collected

Depends on the focus of the encounter (i.e., prenatal, pediatric, and counseling). As a rule, genetics clinics collect a core set of information for each patient, including demographic data and family medical history. A detailed physical exam and diagnosis, if known, as well as a case summary, is also usually available.

Copies of outpatient diagnostic tests and procedures may also be found. Clinics may use multiple disease classification systems depending on the diagnosis (e.g., ICD-9-CM, ISCN or *International System for Human Cytogenetic Nomenclature*, [Mitelman, 1995]) and/or use proprietary coding systems (e.g., POSSUM, Mendelian Inheritance in Man). Clinic charts may also include letters and notes from other physicians, results of research studies, or diagnostic testing that borders on research.

Accessibility and Retrievability

Usually the medical charts for clients/patients are available at the clinic site for review and abstraction. Many clinics collect information in database format for insurance purposes, clinic needs, and network-wide data collection. Due to the nature of the information gathered, the data often are retained permanently. However, state statutes should be consulted for statute of limitations for health information.

Strengths as a Data Source

- *Accuracy.* High quality. The status of a diagnosis is qualified (i.e., the definite, rule out, possible). Although some patients never get a definitive diagnosis, the differential diagnosis is usually provided.
- *Level of detail.* High quality. Specific information on syndromes (identification and description of dysmorphic features) and chromosomal anomalies is often provided.
- *Case identification.* Specialty clinics, like those for genetics, are important outpatient data sources. Previously unknown cases may be identified for the surveillance program.
- *Case identification or screening.* This is a useful source for prenatal diagnosis cases. Clinics may provide diagnosis and/or genetic counseling services.
- *Retrievability.* Most pertinent information is entered into an electronic file (i.e., a database). This facilitates requesting specific pieces of information that can be extracted in electronic format.

Weaknesses as a Data Source

- *Population base.* May not be well defined.
- *Incomplete information.* Nature of the clinic business or the clinic encounter determines whether the complete diagnostic picture is available (i.e., the case may be referred for cytogenetics laboratory confirmation only).
- *Timeliness of diagnosis.* Some diagnoses are not confirmed until multiple diagnostic procedures have been conducted. Some syndromes take a long time to be diagnosed definitively.
- *Follow-up*. Often a case is referred for consultation and is lost to future tracking. This is important if the diagnosis is reported to the surveillance program as possible or rule out and is in the continuing or discovery phase.

Liaisons and Partnerships

- *Genetic counselors.* Clinics are often staffed by genetic counselors who contribute documentation concerning a patient's evaluation. They are often accessible to surveillance staff if a medical records review or other follow-up is needed.
- *Database managers and other office administrators at clinic sites.* Clinical information is often abstracted from documentation in the medical record for billing, research, or other clinic use. These persons can assist the surveillance staff in identifying efficient reporting and case identification methods.
- *Network system managers.* Regional genetics information may be collected and compiled in a database. Like hospital discharge data, regional genetics information is collected from participating clinics in a standardized format and compiled in a centralized format. Surveillance staff can utilize the efficiency of accessing a centralized database and bypass having to collect the case reports from individual clinics. Of importance is the fact that data from these sources are unlikely to include personal identifiers

Issues to Consider

- *Scope of information collected*. Genetics clinics may collect information and provide a diagnosis that extends beyond the types of defects included in a birth defects surveillance system. Passive case ascertainment systems should be precise in specifying the diagnoses that are included in the program's case definition and which are reportable. Active case ascertainment programs could improve efficiencies by developing a more precise list of diagnoses and medical conditions that can be used to screen for potential birth defects cases in the database or log of the clinic.
- *Confidentiality issues.* Genetics information may be protected by additional federal or state statutes. The surveillance system should research applicable legislation, and if necessary, strengthen security procedures and processes in the surveillance system.

References

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