The National Organization for Rare Disorders (NORD) Orphan Diseases and Orphan Drugs

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The National Organization for Rare Disorders (NORD) is a national non-profit health agency dedicated to the prevention, control and new treatments for rare "orphan diseases." NORD was formed by a coalition of voluntary health agencies and individuals with rare disorders who worked together for passage of the Orphan Drug Act of 1983. This federal law provided financial incentives to entice pharmaceutical manufacturers into developing new treatments for small populations of patients ("orphan drugs").

Under the law an "orphan disease" is one which afflicts fewer than 200,000 Americans. Drugs to treat larger numbers of people can also be designated as orphan drugs if the manufacturer can prove that there is no reasonable expectation that profits from the sale of the drug will outweigh the cost of developing the compound through FDA approval. Drugs that fall into this category would be unpatentable compounds, or drugs to treat a subcategory of patients with a prevalent disease (e.g., sub-types of epilepsy).

NORD is a clearinghouse for information about rare disorders and orphan drugs. Medical information about orphan diseases is often difficult to retrieve, outdated or too complicated for the layman to understand. In response to this need, NORD operates the Rare Disease Database (RDB) on CompuServe, the nation's largest electronic information system. RDB is accessible through a personal computer with modem. Each disease entry is written in language understandable to patients and provides referrals to support groups, clinics, researchers or resources that one may contact for further in-depth information. Both standard and investigational therapies are listed for each disease so that doctors and patients can learn about the availability of new orphan drugs when they exist.

To date, the FDA has designated over 250 orphan drugs; 33 of those compounds are approved for marketing. Some orphan drugs which have been developed by academic scientists may never be approved by the FDA even though they are proven safe and effective, simply because they continue to have no commercial sponsor.

A good example of an important orphan therapy is cysteamine, for treatment of the hereditary and once fatal kidney disease cystinosis. Children with cystinosis invariably died at a young age unless they underwent dialysis and eventually kidney transplantation. The outcome even after these procedures was not very positive because cystinosis is an inborn error of metabolism, and the underlying metabolic problem could not be corrected through dialysis or transplant.

Cysteamine was developed by academic scientists, and it enables cystinosis patients to avoid severe kidney damage. Since the drug has no commercial sponsor, it will continue to be an investigational drug until a pharmaceutical manufacturer adopts it and applies for marketing approval. The cost of treating 100 children with cysteamine for one year is approximately $50,000; the cost of dialysis and/or transplant for these children would be approximately $5 million for one year.

Another example of an important orphan drug is L-5HTP, which was also developed in academia for treatment of postanoxic myoclonus. This drug was adopted by a small generic manufacturer who is not experienced in FDA's approval process for new drugs. The company has pledged to provide the compound for free to any patient who needs it; therefore, in the future L-5HTP will remain as an investigational drug.

It is important to realize that many orphan drugs will continue to remain in limbo; although they are safe and effective they may never reach the marketplace even though they will be available to patients under the FDA's Treatment IND or Open Protocol provisions of the Orphan Drug Act. In recent years the health insurance industry has increasingly denied reimbursement for experimental therapies and related medical costs, whereas people with orphan diseases often have no other treatment options. The fact is, many orphan diseases that were untreatable only a few years ago are now treatable because orphan therapies are available to alleviate the illness. Insurers and underwriters should stay abreast of orphan drug development in order to understand the implications of treatment advances in these little-known illnesses.

There are over 5,000 orphan diseases affecting approximately 20 million Americans. Each illness affects fewer than 200,000 people in the U.S. Since there are 257 designated orphan drugs, it is clear that major strides will have to be made in coming years to truly affect the orphan drug/orphan disease problem. The congressionally mandated National Commission on Orphan Diseases is due to submit a report to Congress during February 1989 with recommendations that may speed the understanding and alleviation of orphan diseases. It is NORD's role to assure that the benefits of biomedical research, pharmaceutical development and efforts of the government and health related industries reach the 20 million patients whose lives are touched by orphan diseases.

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