

CLINICAL AND NEUROPHYSIOLOGICAL FEATURES OF NEUROPATHY OF THE FACIAL NERVE

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ABSTRACT. Biallelic mutations in the PTRH2 gene have been associated with infantile multisystem neurological, endocrine, and pancreatic disease (IMNEPD), a rare autosomal recessive disorder of variable expressivity characterized by global developmental delay, intellectual disability or borderline IQ level, sensorineural hearing loss, ataxia, and pancreatic insufficiency. Various additional features may be included, such as peripheral neuropathy, facial dysmorphism, hypothyroidism, hepatic fibrosis, postnatal microcephaly, cerebellar atrophy, and epilepsy. Here, we report the first Italian family presenting only predominant neurological features.

The chapter is organized in three sections. First, it outlines the mechanisms by which the Committee postulates that health insurance affects health-related outcomes. Whether one has health insurance, a regular source of care and, if one is uninsured, the length of time that one is without coverage all influence access to care and affect health-related outcomes.

The second section discusses issues related to the measurement of health insurance effects and considerations of research design that affect the inferences that can be drawn. It explores analytic strategies to distinguish the effects of health insurance status from those of personal attributes that are correlated with health insurance, including health status, race and ethnicity, and socioeconomic status, which may confound the results of studies that relate health insurance to health outcomes. The section gives particular attention to the two-way causal relation ship between health status and insurance status.² It also describes the major population surveys and databases that provide information about Americans' use of health care and epidemiological information about health status and disease prevalence. These sources provide the data for many of the most informative studies reviewed.

Methods

Extensive neurological and neurophysiological evaluations have been conducted on the two affected brothers and their healthy mother since 1996. The diagnosis of peripheral neuropathy of probable hereditary origin was confirmed through a sural nerve biopsy. Exome sequencing was performed after the analysis of major neuropathy-associated genes yielded negative results.

Health insurance facilitates access to health care by removing or diminishing financial barriers to obtaining care. Among people who have insurance, the extent of cost sharing also influences the use of health care (Newhouse et al., 1993; Zweifel and Manning, 2000). An extensive body of research consistently finds a strong and positive relationship between health insurance and access to care, even as the definitions and measures of access have been strengthened. Population-based surveys conducted over the past three decades have evaluated access to primary care in relation to health insurance status with measures such as any physician visit within a year, the number of physician visits per year, having a regular source of care, and the ability to obtain care when needed (Freeman and Corey, 1993; Hafner-Eaton, 1993; Newacheck et al., 1998; Nelson et al., 1999; Zuvekas and Weinick, 1999; Haley and Zuckerman, 2000; Kasper et al., 2000; Shi, 2000; Weinick et al., 2000; Hoffman et al., 2001).

Public policy and health care industry interests in high-quality and efficient health care have developed in tandem with the progress of clinical effectiveness research over the past decade. The standards of evidence for the efficacy of health insurance in promoting better health outcomes have evolved from enumerating physician visits to measurable improvements in effective processes of care. The notion of “access” itself has shifted from a simple measure of utilization to measures that incorporate the quality of care and health outcomes. In 1993, the Institute of Medicine (IOM) Committee on Monitoring Access to Personal Health Care Services reconceptualized access as “the timely use of personal health services to achieve the best possible health outcomes” and recommended a set of health outcome measures that could serve to monitor populations over time for access to basic health services (Millman, 1993).

Results

Whole-exome sequencing analysis identified the homozygous substitution c.256C>T (p.Gln86Ter) in the *PTRH2* gene in the two siblings. According to American College of Medical Genetics and Genomics (ACMG) guidelines, the variant has been classified as pathogenic.

At 48 years old, the proband's reevaluation confirmed a demyelinating sensorimotor polyneuropathy with bilateral sensorineural hearing loss that had been noted since he was 13. Additionally, drug-resistant epileptic seizures occurred when he was 32 years old. No hepatic or endocrinological signs developed. The younger affected brother, 47 years old, has an overlapping clinical presentation without epilepsy.

In studies of access to care and health outcomes, several factors mediate the relationship between health insurance and health-related outcomes. These include being able to see a provider when one believes care is needed, having a regular source of health care, having continuity of coverage, and the duration of periods without health

insurance. Measures for each of these factors provide some information about an individual's or population's access to health services that supplements the measurement of health insurance status at a given point in time. These measures are discussed below.

The ability to see a physician or other health care provider when one believes medical attention is needed is a fundamental and intuitive measure of access to health care. Most Americans mistakenly believe that people without health insurance have this level of access (IOM, 2001a). Although the lack of health insurance is not the only reason someone might not be able to see a health care practitioner when needed, it is a major one.³ Adults without health insurance are far more likely to go without health care that they believe they need than are adults with health insurance of any kind (Lurie et al., 1984, 1986; Berk and Schur, 1998; Burstin et al., 1998; Baker et al., 2000; Kasper et al., 2000; Schoen and DesRoches, 2000; Davidoff et al., 2001; Holahan and Spillman, 2002).

Interpretation

Our findings expand the clinical phenotype and further demonstrate the clinical heterogeneity related to *PTRH2* variants. We thereby hope to better define IMNEPD and facilitate the identification and diagnosis of this novel disease entity.

While the overall percentage of adults who reported that cost prevented them from seeing a doctor in the previous 12 months increased only slightly from 10 percent to 11 percent between 1991 and 1996, the proportion of uninsured adults who reported this barrier to care increased from 28 to 35 percent, and the fraction of insured adults reporting this barrier decreased slightly from 8 to 7 percent (Nelson et al., 1999).⁴ In 1998, nearly 70 percent of uninsured adults in poor health could not see a doctor at some time during the year because of cost (Ayanian et al., 2000). A study that polled 1,100 patients four months after their initial visit to an emergency department found that patients who lost their health insurance were more than twice as likely as those who maintained their coverage to have delayed seeking care in the four-month interval (Burstin et al., 1998).

Evaluations based on professional judgment confirm findings based on a subjectively determined need for care. In one study with a national probability sample of almost 3,500 adult respondents, a physician panel identified 15 serious conditions for which they deemed medical attention necessary (Baker et al., 2000). In an analysis that adjusted for demographic and economic characteristics and also for health status and having a regular source of care, the authors found that an uninsured adult was much less likely than an insured adult to get care for a reported symptom (odds ratio [OR] = 0.43). Examining only those symptoms for which the respondent thought care was needed, those without insurance were even less likely to have received care (OR = 0.28). Among those who did not receive needed care, the uninsured were far more

likely than those with insurance to report that they did not get care because of cost (95 percent and 23 percent, respectively) (Baker et al., 2000).⁵

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