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Needs reassessment of staffing and time management in clinical genetics

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Innovative practice models that improve efficiency or justify additional staffing may be needed to help clinical geneticists cope with increasing workloads, according to the results of an international survey reported in this issue. The results revealed that between 25% and 42% of a clinician's time is taken up with face-to-face patient interaction. The remainder of the time is divided among various administrative tasks, including research on the increasingly complex array of tests and genomic databases, which are rapidly evolving. Time spent with patients now requires explanations of complex topics, such as depth of coverage, inclusiveness, testing of genes for which there may be no evidence-based management recommendations, and the probability of finding variants of uncertain significance, as reported by the survey research team, who are based in Israel and Canada. Administered online to attendees of professional clinical genetics meetings, the survey elicited responses from 151 professionals, mainly from Europe, North America, and the Middle East.

On average, survey respondents reported about 17 patient encounters per week, including both initial and follow-up visits. The study authors conclude that current work conditions are not conducive to a sustainable future, given that "it is unlikely that the direct reimbursement of genetics services can cover such high costs." They argue that as the influence of genetics on medicine increases, healthcare institutions should consider the impact of offering high-level genetics services on overall goals of offering high quality of care, research, and academic performance. *—Karyn Hede, News Editor*

Long-term study of familial hypercholesterolemia expands range of genetic defects

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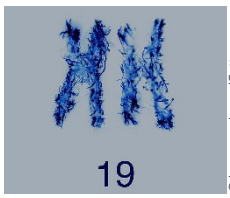

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A 15-year study of familial hypercholesterolemia (FH) in Portugal has led to identification of a wide variety of mutations in several genes. Because FH, an autosomal dominant disorder of lipid metabolism, can be treated to reduce the risk of life-threatening cardiovascular events, universal screening is recommended by the World Health Organization and has been conducted in Portugal since 1999. Long-term research led the study investigators to conclude that molecular analysis of FH patients should be expanded to include all coding sequence, promoter, and splice-site regions for four genes—*LDLR*, *PCSK9*, *APOB*, and *APOE*—due to the recently discovered mutations. The research project included 2,122 people diagnosed with FH. Over the course of the study, 13 novel variants of *APOB*, whose protein product is the main component of low-density lipoproteins, were identified in the study group. Two of these are disease-causing variants, four are polymorphisms, and seven are rare variants requiring further study. Three of the variants are reported in this issue for the first time. Study investigators are currently conducting functional analysis of the novel variants and developing a gene panel for clinical use. Additionally, the authors suggest that a more cost-effective approach to FH identification needs to be established, as well as a case-finding program and a cascade screening program. *—Karyn Hede, News Editor*

NEWS BRIEFS

Endangered Y chromosome may be disposable

Courtesy of Yasuhiro Yamauchi

The Y chromosome, that bastion of masculinity, may disappear one day and not be missed. Studies suggest that the Y chromosome has been shrinking rapidly on an evolutionary time scale. Now a University of Hawaii–based research team, as reported in the 29 January 2016 issue of *Science*, has succeeded in experimentally eliminating the chromosome while maintaining fertile male mice. Only two genes from the Y chromosome appear to be critical: *SRY*, which initiates production of testes, and *Eif2s3y*, which initiates production of sperm. The other genes on the Y chromosome can apparently be dispensed with, at least for reproductive purposes. In fact, the research team substituted the entire Y chromosome with an X chromosome and then overexpressed two non-Y residing genes next in line in the genetic cascade that leads to testes and sperm. That was enough to produce male mice with sperm capable of fertilizing a mouse egg. However, because the sperm produced by the Y-less mice were defective, with no functional tails, investigators used *in vitro* fertilization (IVF) to see if they could successfully reproduce. The IVF worked, but male progeny were infertile, while females could reproduce normally. The experiment raises the question of what all those other genes on the Y chromosome are good for if only