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A self-guided tour of one's genome

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Debate within the clinical genetics community about how to return whole-genome and whole-exome data to individuals has tended to focus on genomic data as "results." In this issue, a research team from the University of Washington proposes shifting the emphasis from returning results to providing a kind of atlas that can be accessed over time as health and life circumstances warrant. This genomic atlas would, in their view, allow a self-directed tour that could be as short or as long a journey as desired. Reframing the debate to focus on self-guided management rather than return of results would allow individuals to determine whether and when they receive results. It would also, the authors argue, "reduce the practical burden of forcing providers to act as gatekeepers of the broad scope of results with ever-changing implications for the health of their patients." In such a system, the roles of genetic counselors and clinical geneticists would, over time, shift to providing an interpretive safety net for recipients. Return of results would no longer be a one-time interaction but an ongoing process. Genetic counseling would become a long-term relationship at a pace driven by recipients. The authors suggest that this platform would relieve some of the ethical burden currently felt by clinicians while also conforming to standards of bioethics. However, the question of who should bear the responsibility of maintaining sequence data remains. Within the proposed framework, the authors propose that researchers focus on "how, by what mechanisms, to what extent, in what contexts, and with what outcomes results will be offered and returned." *—Karyn Hede, News Editor*

The hereditary ataxias: a review

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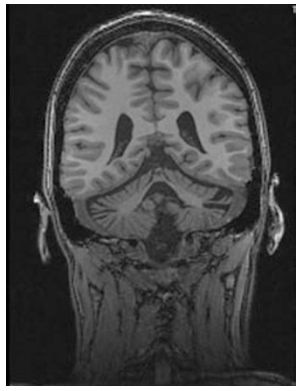
The hereditary ataxias often present with an array of overlapping symptoms that require molecular genetic testing to discriminate between them. All individuals with ataxia have problems with gross motor coordination due to defects in the nervous system that affect movement and balance. In addition, they may develop problems with eye movement, spasticity, neuropathy, and cognitive/behavioral difficulties. Establishing the specific molecular cause of hereditary ataxia for any given individual generally requires obtaining medical and family history, physical and neurologic examinations, and neuroimaging, in addition to molecular genetic testing. In this issue, Jayadev and Bird present the current understanding of these diverse disorders, including both the autosomal dominant and recessive forms, and review accepted steps for differential diagnosis. The authors review evaluation strategies, including non-DNA and DNA testing methods, as well as available prenatal testing for some forms of the disorder. Progress in identifying causative genes has enabled diagnosis of an estimated 50–60% of the dominant hereditary ataxias via a battery of genetic tests. Many laboratories first test for the genes associated with the more common ataxias (*SCA1*, *SCA2*, *SCA3*, *SCA6*, and *SCA7*), offering a second tier of tests only if the first group yields no positive diagnosis. Challenges and approaches to genetic counseling and disease management are also briefly covered. *—Karyn Hede, News Editor*



EDITORIAL

Visit the new Smithsonian genome exhibit

Don Hurlbert, Smithsonian Institution

We live in an age in which science and technology dominate every facet of our lives. From genomics to informatics, science matters. And yet the general public, our political leaders, and those who make important policy decisions all too frequently lack the most basic understanding of how our world works.

However, education is about much more than the imparting of facts and knowledge. Indeed, I would argue that the first job of an educator is to inspire and excite his or her audience—whether that audience is an elementary school classroom, medical students, or the public. Once a potential student has been captivated by a subject, the rest comes naturally as they actively seek further knowledge.

It is in part this aspect of education—the need to inspire—that makes our nation's museums vital to an informed public. Now, the venerable Smithsonian National Museum of Natural History, in Washington, DC, is using its formidable power to inspire in the service of genomics education. On 14 June 2013, a new exhibit opened: "Genome: Unlocking Life's Code," cohosted by the National Human Genome Research Institute. Visitors learn about the power and beauty of genomics and explore, in a variety of interactive ways, multiple facets of the genome. They get a sense of the immense size of the genome, the analytical chal-

EDITORIAL *(continued)*

lenges with which we are struggling, the potential for its application in medicine, and the light it sheds on who we are at the most fundamental level.

Generations of scientists have been inspired by their visits to museums where they first glimpsed the power and beauty of science. The sheer beauty of DNA and the human genome, coupled with the mastery of the Smithsonian's presentation, will inspire future generations about a subject important to society's future and dear to the heart of the readers of this journal.

"Genome: Unlocking Life's Code" will be open at the Smithsonian National Museum of Natural History through August 2014, after which it will travel throughout the United States. Plan to visit with your family!

—James P. Evans, Editor-in-Chief

Genetics in Medicine | Mission Statement

Genetics in Medicine is a monthly journal committed to the timely publication of:

- Original reports which enhance the knowledge and practice of medical genetics
- Strategies and innovative approaches to the education of medical providers at all levels in the realm of genetics

As the official journal of the American College of Medical Genetics and Genomics (ACMG), the journal will:

- Provide a forum for discussion, debate and innovation concerning the changing and expanding role of medical genetics within the broader context of medicine
- Fulfill our responsibility to the College membership through the publication of guidelines, policy statements and other information that enhances the practice and understanding of medical genetics

Finally, as genetics becomes increasingly important in the wider medical arena, we will be an accessible and authoritative resource for the dissemination of medical genetic knowledge to providers outside of the genetics community through appropriate reviews, discussions, recommendations and guidelines.