

## Clinical genetics provider real-time workflow study

### To the Editor:

I read with interest the article on real-time workflow study by McPherson et al.<sup>1</sup> The authors are to be commended for this timely and valuable study. Also noteworthy is that this new study yielded results quite comparable with similar investigations 20 years ago.<sup>2,3</sup> My point of concern is their study design in patient type breakdown. Their patient types were new visits, prenatal, and follow-up. In a previous study by Cooksey et al.,<sup>4</sup> the patient types included general (36%), pediatric (28%), reproductive (15%), metabolic (14%), and adult (7%). However, with the advance of molecular genetics that has been translated into clinical practice at a phenomenal pace, the number of DNA-based molecular testing has grown roughly 1200% from 1997 to 2007.<sup>5</sup> As a consequence, the percentage of adult cases has increased more significantly than other patient types. At our center, they comprise ~50% of our total number of patients. Therefore, when documenting the workload of a clinical genetics unit, it is increasingly relevant to make the distinction between those cases that are diagnostic conundrums of multiple organ-system disorders and those with a clear diagnosis, often adult-onset, of single, or primarily single, organ-system involvement. Examples of the latter category include hereditary cancers, familial hypertrophic cardiomyopathies, dysrhythmias, thrombophilia, and polycystic kidney diseases. These disorders characteristically fit into a pediatric or adult medicine subspecialist's realm of practice and are oftentimes diagnosed by these subspecialists. The patient and their family members come to genetics primarily for genetic counseling. The former category, on the other hand, typically, although not exclusively, consists of patients with multiorgan-system involvement that defy compartmentalized approaches of the subspecialists, but the case complexity is beyond the scope of practice of the general practitioners. Conditions in this category often carry a syndromic diagnosis of a rare disorder such as achondrogenesis, Robinow syndrome, or mitochondrial respiratory chain disorder. In many cases, the diagnosis remains elusive despite comprehensive workups. I find that the MD geneticist's time spent on patient-care-related activities, both direct and indirect, for these categories differs far more significantly than new versus follow-up visits of the same category, and this is true for different genetics centers that I have worked at. For complex cases, an MD geneticist typically can see only 2–4 patients in half a day clinic plus substantial pre- and postclinic time on literature search, database search, review of outside medical records, dictation of detailed clinic letters, and ongoing review of consult notes, imaging results and other investigations. It is also not uncommon for these cases to be written up for publications, thus blurring the division of patient-care-related activities and academic activities. For the former category, however, an MD geneticist can supervise a number of concurrent genetic counselors' (GC) clinics and see up to 12–16 patients in a half day with only minimum pre- and postclinic time, as the bulk of the work, including clinic letters to the referring physicians, is

done by the counselors. A third category in our practice is a GC-MD joint clinic where cases that do not fit into either of the above categories are seen. These often include cases with a known diagnosis such as a chromosomal anomaly where a genetic assessment is yet to be done. Genetic counseling and MD assessment happen in the same clinic, and the time division is roughly 2/3 and 1/3 for GC and MD, respectively for both direct and indirect patient care activities. Furthermore, most MD geneticists do not manage a patient's day-to-day medical issues.<sup>5</sup> Consequently, the number of follow-up visits is limited. Typically, more new patients are seen each year than at follow-ups.<sup>5</sup>

In light of the dynamic shift in the scope and content of medical genetics practice, categorizing patient type according to the nature of the encounter is becoming more relevant than dividing them into new versus follow-up visits when workflow and workload are being assessed.

**Chumei Li, MD, PhD**

*Clinical Genetics Program, McMaster Children's Hospital  
McMaster University Medical Center, Hamilton  
Ontario, Canada*

### References

1. McPherson E, Zaleski C, Benishek K, et al. Clinical genetics provider real-time workflow study. *Genet Med* 2008;10:699–706.
2. Bernhardt BA, Pyeritz RE. The economics of clinical genetics services. III. Cognitive genetics services are not self-supporting. *Am J Hum Genet* 1989;44:288–293.
3. Pletcher BA, Jewett EAB, Cull WL, et al. The practice of clinical genet: a survey of practitioners. *Genet Med* 2002;4:142–149.
4. Cooksey JA, Forte G, Flanagan PA, et al. The medical genetics workforce: an analysis of clinical geneticist subgroups. *Genet Med* 2006;8:603–614.
5. GeneReview. Growth of laboratory directory. Available at: <http://www.genetests.org>. Accessed 2008.