Reply to Letter from Dr. Chumei Li:

Dr. Li's letter confirms our expectation that, despite the similarity of our results to those of similar investigations 20 years ago, there will always be differences between centers. In addition to our breakdown by visit type (new, prenatal, and follow-up), we do provide some information on patient type in Table 2.[1] We document that one third of our new visits are for cancer, more than one fourth of our new patients have previously undiagnosed multiple congenital anomalies, and another one fourth have other unknown or incompletely diagnosed conditions. This latter category includes some complex diagnostic problems, such as metabolic or neurodegenerative disorders, and more straightforward issues like Ehlers-Danlos or skeletal dysplasias. Even the latter, however, require considerable time from both a physician and genetic counselor when a new diagnosis is made.

Like Dr. Li, we have seen an increase in adult patients who now comprise about half of our new visits. We have found, however, that even those referred for a specific reason, such as cancer or cardiomyopathy, may present significant diagnostic challenges. Our patients rarely have molecular genetic testing before referral, and part of our responsibility is to determine which tests are indicated. Some of the cancer patients have

complex family histories and are offered testing for several different cancer predisposition syndromes. Dilated cardiomyopathy, which requires consideration of muscular dystrophy, mitochondrial, and metabolic disorders, is more frequent than hypertrophic cardiomyopathy in our clinic population. We find that the distinction between complex and simple cases often becomes blurred, for example, if the referring diagnosis turns out to be incorrect or if the family history (which we take during the clinic visit) reveals additional issues for which evaluation and counseling are needed.

As mentioned in our article, we usually schedule a 2-hour time block for new patients. This includes both genetic counselor and physician time, although for known diagnoses (such as cancer), the physician may be supervising two counselors simultaneously. Because of requirements for supervision, and because at our center genetic counselors are unable to bill for their services unless the physician is in the room, the genetic counselor and physician always work together for at least part of the session. The actual allocation of the face-to-face time varies with the diagnosis. As Dr. Li observes, more physician time is required for the complex undiagnosed cases. For a new multiple congenital anomaly patient, both the physician and genetic counselor may spend nearly the full 2 hours in the room with the patient, whereas for straightforward diagnoses, the physician may spend as little as 15 minutes with the patient. We do not, however, have the option for one physician to supervise up to 12–16 patient visits in half a day as suggested by Dr. Li, because our scheduling is limited by the availability of genetic counselors. Furthermore, referral patterns, patient preference, and institutional regulations make it necessary for us to distribute patients over the entire work week. Our practice is most similar to the genetic counselor-MD clinic mentioned in Dr. Li's letter. In fact, for direct and indirect patient care combined, the proportion of genetic counselor to MD time reported in our study (2.3 hr vs. 1.2 hr per 1 hr slot) is very close to the two third to one third reported by Dr. Li for his genetic counselor-MD clinic.

The issues raised by Dr. Li may reflect not only differences between clinics but also differences in practice between the United States and Canada. Much of our indirect patient care time is devoted to insurance authorizations and documentation required for reimbursement. Patient referral patterns, the distribution of work between the primary care physician and the specialist, and scheduling of specialist visits may be dependent on the structure of the health care system.

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Reference

1. McPherson E, Zaleski C, Benishek K, et al. Clinical genetics real-time workflow study. *Genet Med* 2008;10:699–706.