I have always considered preventing future health problems and targeting the root of illness to be an important part of any physician’s practice, but I did not really understand the term “public health” until my first year of medical school. Once I became aware of public health as a field, I immediately started to think about public health opportunities for the summer between my first and second years of medical school.

Through contacts in Madison, I learned of a summer genetics internship at the Marshfield Clinic, where I could work on a project related to the Wisconsin newborn screening program. Although I was somewhat hesitant to venture up to the Marshfield Clinic for the summer because it was out of my Madison comfort zone, I was intrigued by the promise of an interesting project at a renowned medical center. In the end, this internship was a great lesson in the realities of public health and medicine.

My main task for the summer was to research 22q11 deletion syndrome (22q11DS), otherwise known as DiGeorge or Velocardiofacial syndrome, in order to determine its suitability for addition to Wisconsin’s newborn screening panel. At first glance, it seemed like a perfect candidate for newborn screening. The syndrome affects at least 1 in 5000 babies born, which is comparable to other diseases on the newborn screening panel. It is also associated with severe medical problems, such as cardiac defects, immune deficiency, and hypocalcemic seizures. Identifying newborns who have 22q11DS could allow for earlier treatment to improve outcomes. It might also relieve some of the stress of a “diagnostic odyssey” that families of affected children sometimes experience due to uncertain diagnosis. An inexpensive, accurate PCR (polymerase chain reaction) test was expected to be available soon in order to identify newborns with the gene deletion.

However, as I reviewed the literature and talked with state experts in the fields of newborn screening, genetics, public health, ethics, cardiology, and immunology, it became clear that the situation was much more complex. The syndrome varies widely between individuals; hypocalcemia and immunodeficiency requiring urgent care are seen in a minority of cases. Cardiac defects do occur in about three-quarters of the cases, but there is again quite a variety of manifestations, from ventricular septal defects to interrupted aortic arch. Some of these could be identified on physical exam due to murmurs or cyanosis, so it is not clear that newborn screening would be of significant benefit in all cases. The situation is further complicated by factors including potential negative effects on parent-child bonding, the ethics of newborn screening for a condition including adulthood-presenting features such as mental illness, setting precedent for other newborn screening tests, and lack of proven benefits of early diagnosis. There is also risk for potential incidental findings of unclear significance, such as 22q11 duplication syndrome, a recently recognized microduplication syndrome whose features are not well defined beyond some cases of mental retardation, learning disability, growth retardation, and other problems. It has also been seen in individuals with no identifiable effects, so early diagnosis would likely offer little medical benefit.

Of course, weighing these risks and benefits depends on personal judgments, which vary between individuals. For example, in interviewing patients with 22q11DS and their families over the summer, I found that a majority felt the benefits of screening outweighed the risks, and many professionals I spoke to appeared to feel the same way. However, others, myself

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included, are a bit more hesitant and remain unconvinced that newborn screening will provide clear benefit. Medical technologies are advancing in a way that makes it likely we will soon have technologies to allow early diagnosis of a plethora of genetic diseases. In some cases, these early diagnoses may lead to dramatic benefit, as with diseases like phenylketonuria. However, I now realize that even if screening tests are available, they are not always appropriate, especially when management would not be dramatically altered. The mandated nature of newborn screening prevents families and individuals from making decisions consistent with their personal beliefs. It remains to be seen what decisions will be made at the state level regarding screening for 22q11DS, but in the course of my research it became clear that, as simple as the term “newborn screening” sounds to the general public, it is much more complex and deserves greater consideration than it is usually given.

Even though wrestling with this complex problem was rewarding on its own, leading to two manuscripts, one published in Genetics in Medicine and one under review by the Journal of Genetic Counseling, this research describes only a portion of my summer experience. While working in the Marshfield Clinic’s Medical Genetics department, I had the chance to observe numerous patient appointments, covering genetic issues across the entire life span. I saw how genetics plays a role before birth, in instances of preconception advising and abnormal prenatal screening results. I saw the role of genetics consultations in childhood, including sweat tests for positive cystic fibrosis newborn screens, assessment of children with multiple congenital anomalies, and investigation for a genetic cause of a patient’s autism. I saw, too, how genetics can be important in adulthood, providing guidance and treatment for diseases like Huntington’s disease, hearing loss, and Marfan syndrome. These clinical experiences helped me to understand the role of genetics in primary care medicine in ways that my genetics course did not.

In addition to this clinical knowledge, I also learned important genetics skills that will help me treat patients in my future practice. A pedigree is an important tool for documenting a patient’s detailed family history in a precise, yet expedient manner. After seeing the important information that can be drawn from an accurate pedigree, I’ll never forget to inquire about all the appropriate details, such as half-siblings, Ashkenazi Jewish background, or age of onset of cancer. I am embarrassed to think of the many incomplete family histories I obtained from standardized patients last year, but after this experience, I won’t make that mistake again.

I also acquired a taste of the specialized physical exam skills geneticists use. I never knew that measuring the distance between the eyes, the length of the fingers, or the flexibility of joints could be useful in diagnosis. Perhaps someday I will even be able to identify a genetic syndrome based on “facies” alone, like some clinicians can. If not, I now have a better idea of the diagnostic tests that can help with genetic diagnosis, including microarray, FISH (fluorescence in situ hybridization), and DNA sequencing. Even ultrasound can give valuable information for a prenatal genetic diagnosis.

Overall, this seemingly simple project (the suitability of 22q11 deletion syndrome for newborn screening) blossomed into an unforgettable experience in public health and its intersection with medicine. I’m sure I will carry the lessons of this summer far into my future medical practice.

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