There have been striking changes in genetic testing, from chromosomal microarray technology to next-generation sequencing along with advances in treatment of some genetic disorders. Next-generation sequencing has led to the development of specific multigene panels for groups of disorders to whole exome and genome sequencing. However, it is challenging to know the appropriate testing for respective patients and the limitations of that testing. This is where the clinical geneticist can be critical in the diagnosis and care of the patient. For example, chromosomal microarray and Fragile X DNA testing are still the first genetic testing to perform for a patient with intellectual disability, autism spectrum disorder, and/or multiple congenital anomalies.

This issue of Pediatric Annals is designed to review some specific conditions and principles that are important for the practicing pediatrician, and to give the latest updates in these areas in screening, diagnostic testing, and treatment. The goal of this issue is to give the latest updates in newborn screening, lysosomal storage disorders, microdeletion/microduplication syndromes, pediatric cancer predisposition syndromes, and pharmacogenomics.

In the article, “Updates in Newborn Screening,” Dr. Farrah Rajabi reviews the latest additions to the newborn screen and the rationale for choosing the particular conditions for newborn screening. Newborn screening was initially geared toward treatable inborn errors of metabolism, such as phenylketonuria, and it was biochemically based. It has expanded, detecting hearing loss, immunodeficiencies, congenital heart disease, and more recently lysosomal and peroxisomal disorders. The pediatrician will see positive results, which requires familiarity with the basics of these conditions.

As a companion to the article on newborn screening, the article, “Enzyme Replacement Therapy: A Review and Its Role in Treating Lysosomal Storage Diseases,” by Dr. Mindy Li discusses the various lysosomal disorders (LSDs), their treatment, and the institution of newborn screening for LSDs. With the advances in enzyme replacement therapy, some treatment has become possible for previously untreatable conditions.

Chromosomal microarray technology led to the delineation of multiple microdeletion/microduplication syndromes. Together, these microdeletions/microduplication syndromes account for a significant proportion of patients with intellectual disability, developmental delay, and autism. Therefore, pediatricians should expect to have children who are affected in their practices. In the article, “An Update on Common Chromosome Microdeletion and Microduplication Syndromes,” Dr. Paula Goldenberg reviews specific relatively common microdeletion/microduplication syndromes including 22q11 deletion and duplication syndromes, Williams syndrome, 7q11.23 duplication syndrome, Smith Magenis syndromes, Potocki-Lupski syndrome and 16p deletion, and duplication syndromes delineating the major issues of each of these conditions. One important component of this article is the description of the deletions and duplications of the same chromosomal region and how they may differ in their features and management.

The underlying genetics of pediatric cancer is reviewed, which may have implications for treatment, monitoring, and genetic counseling for the families. Although cancer is relatively rare in pediatrics, there is a large genetic component to these cancers. With advances in genetic testing technology, identification of the underlying genetic etiology has become more common. The article, “Recognizing and Managing Children with a Pediatric Cancer Predisposition Syndrome: A Guide for the Pediatrician,” by Stephanie A. Coury and her colleagues reviews the common pediatric cancers and the warning signs of cancer for the pediatrician to note. It also reviews the indicators for a genetic cancer syndrome and presents a tool for such identification.

In the final article, “Pharmacogenomic Primer for the Pediatrician—What Is Real-
ly Important?,” by Dr. Shannon Manzi, the emerging field of pharmacogenomics and how it relates to pediatrics is discussed. The area of pharmacogenomics is not one with which practicing pediatricians may be familiar. It is not used routinely in pediatric practice, but it is important in certain situations. However, with expanded precision or personalized medicine, pharmacogenomics may become more relevant and common. Dr. Manzi reviews the general principles of pharmacogenomics and when testing for pharmacogenetic variants is indicated and may be useful, such as in the treatment of cystic fibrosis and in oncology.

REFERENCES

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