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Joint clinical report updates guidance on neurofibromatosis type 1

by David T. Miller M.D., Ph.D., FACMG, FAAP; Debra L. Freedenberg M.D., Ph.D., M.Phil., FACMG, FAAP

Every pediatrician likely has encountered at least one patient with neurofibromatosis type 1 (NF1) but may not be aware of current care recommendations for these children.

NF1 is one of the most common single-gene conditions, with a prevalence of about one in 3,000 people, similar to cystic fibrosis.

NF1 can cause a wide variety of complications, some of which may not be detectable without dedicated surveillance. Importantly, catching these complications early can affect outcomes.

An updated AAP clinical report will be a valuable resource to improve awareness of NF1 care guidance for general pediatricians, geneticists, child neurologists and dermatologists who might play a role in diagnosing or treating children with NF1.

The report, *Health Supervision for Children With Neurofibromatosis Type 1,*from the AAP Council on Genetics and the American College of Medical Genetics and Genomics, is available at https://doi.org/10.1542/peds.2019-0660 and will be published in the May issue of *Pediatrics*.

Since the clinical report was first published in 2008, there have been important developments in diagnosis and treatment that are reflected in this report.

The updated guidance represents a consensus among an interdisciplinary group of experts in the care of individuals with NF1. Several members of the group have served on the Clinical Care Advisory Board of the Children's Tumor Foundation, an organization that sponsors NF research and clinical care.

Diagnosis, treatment

The first step is identifying children who may have NF1.

Symptoms can develop gradually, potentially leading to a delay in diagnosis. Children under age 6 years are at the highest risk of developing an optic pathway glioma that could affect their vision, underscoring the importance of early diagnosis.

With a keen awareness that every primary care physician encounters children with café au lait macules but may lack the experience to determine if these indicate NF1, the report provides a differential diagnosis of multiple café au lait macules. In some cases, making a definitive diagnosis can be difficult, and genetic testing may be considered.

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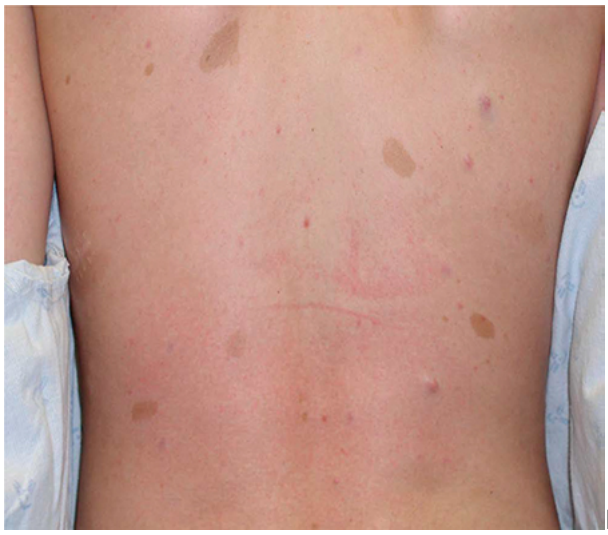
Note multiple cafe au lait macules. A definitive diagnosis of neurofibromatosis can be difficult if only pigmentary symptoms are present, and genetic testing may be considered.

The report discusses the role of genetic testing, which has gained importance due to the recognition of newly described conditions with symptoms that mimic NF1, namely Legius syndrome. Also addressed are emerging trends in genotype-phenotype correlation for patients with NF1.

Another important aspect of this update is a discussion of emerging therapies and symptom-based guidance. Previously, limited therapeutic interventions were available for most genetic syndromes, including NF1. With increased understanding of the underlying cellular pathophysiology, targeted therapies for treatment of some of the more severe manifestations of NF1 are being investigated, such as Ras signaling pathway modulators. A variety of clinical trials, many of them coordinated through the NF Clinical Trials Consortium, are providing evidence for meaningful therapeutic interventions for various features of NF1.

Care coordination

The pediatrician's role as the medical home, providing both care coordination and surveillance, continues to be of critical importance in the management of children with NF1.

While some patients with NF1 need extensive care coordination, the majority of children will have mild manifestations in childhood. With the rapid advances in NF1 management, children can benefit from evaluation at a specialized center with neurofibromatosis expertise working in concert with the medical home.

Drs. Miller and Freedenberg are lead authors of the clinical report. Dr. Freedenberg is a member of the AAP Council on Genetics and a former member of its executive committee.

Resources

- Children's Tumor Foundation
- Neurofibromatosis Network
- American College of Medical Genetics and Genomics