Since January 2020 Elsevier has created a COVID-19 resource centre with free information in English and Mandarin on the novel coronavirus COVID-19. The COVID-19 resource centre is hosted on Elsevier Connect, the company's public news and information website.

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Commentary

As an unfortunate consequence of COVID-19, Issue 2 of Genetics 2022 was postponed. Like many aspects of healthcare, COVID overwhelmed other priorities; however, pediatric and adult dermatologic care continues despite the COVID-19 pandemic, and cutting-edge genetics now returns to our consciousness. Some aspects of the health system that evolved during COVID-19, specifically telehealth, have emerged as an important way to evaluate children with genetic conditions. These children sometimes travel great distances to see sub-specialists, while dealing with complex medical and developmental disabilities that limit their mobility and strain caretakers. Telehealth, while born out of necessity, is likely to remain part of the care of complex genetic patients, allowing patients to reach specialized practitioners near and far. Nevertheless, many patients with genetic disorders are behind on their care and this update will help the practitioner improve their genetic knowledge and ability to manage complex patients.

Current concepts

In the first issue complex genetic disorders were tackled, with many more deep insights into disease-state, work-up and therapeutics in this issue.[1] One study characterizes the current, cutting-edge approach to vascular malformations, simplifying an incredibly complex topic.[2] Although diagnosis has historically rested on clinical and radiographic features, techniques such as next-generation genetic sequencing and droplet digital polymerase chain rection (PCR) can now establish genetic diagnoses even in mosaic conditions. The integration of genetic techniques into the care of vascular malformations allows patients to access targeted genetic therapies, no longer playing a guessing game of phenotype to genotype correlation.[2]

Another study reviews the differentiation of disorders associated with pigmentary mosaicism through categorizations including internal features, and genetic testing, using methods such as whole-exome sequencing and sensitive microarray-based techniques.[3] This wonderful guide to the care of pigmentary mosaicism patients simplifies an abstruse topic, providing the reader with actionable direction down to the best biopsy technique.[3]

Another study reviews the many syndromes associated with both atypical nevi, multiple benign appearing nevi, and nevi whose first appearance is unknown.[4] Understanding the molecular basis of these conditions can aid in the early identification of patients with genetic risk of melanoma or internal malignancies and will hopefully allow for future gene therapy.[4]

Another paper reviews the technological advances that now allow tape stripping to provide predictive value in the diagnosis of melanoma, in addition to its utility in studying gene upregulation and inflammatory mediators in conditions like atopic dermatitis that have complex genetics.[5] This technology, able to be accomplished in person or remotely adds to our armamentarium of diagnostic tools.[5]

Pediatric autoimmunity is associated with a multifactorial myriad of genetic mutations and polymorphisms contributing to the development of the common autoimmune diseases-vitiligo and alopecia areata. In particular, specific genes predisposing to vitiligo and alopecia areata are involved in the pathogenesis of other autoimmune conditions, creating patterned associations and identifying targets for therapeutic intervention.[5]

Another study reviews the genetics of neurocutaneous disorders highlighting how genetic pathways can provide the rationale for newer therapies under development.[6] The model of neurocutaneous disorders serves as a good model for well-understood interdisciplinary care patterns.[7] Additional new ideas in genetics are the engineering of phage therapy to treat genetic diseases associated with chronic infections, including Netherton syndrome.[8] Phage therapy is being developed, however, through genetic engineering and screening of phage genomes, for the treatment of common conditions such as acne.[8] We are sure to hear quite a lot more in the near future in regards to this therapy.

Our final contribution highlights the bread and butter aspects of genodermatoses. Another study reminds us that exceptional care begins with clinical insight and relies on durable surveillance over time.[9] They provide a comprehensive overview of nail changes in patients with congenital

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ichthyosis that highlight features not well-described in the literature.

The future

Now that we can welcome patients back to the office, despite the masks, it behooves us to undress our suspected genetic syndrome patients and look for stigmata, biopsy judiciously, send blood tests (eg, enzyme levels) where needed and make use of the incredible genetic testing options. Genetic options open up so many worlds for our patients which include: provision of excellence in personal care that comes with confirmation of diagnosis, genetic screening for family members in the setting of single-gene disorders, and the emerging field of targeted medical therapies. The future is coming into focus and the combination of modern diagnostic techniques and expanding targeted therapies will revolutionize the care of genetic disorders.

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