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A next-gen sequencing method for tricky spinal muscular atrophy carrier screen

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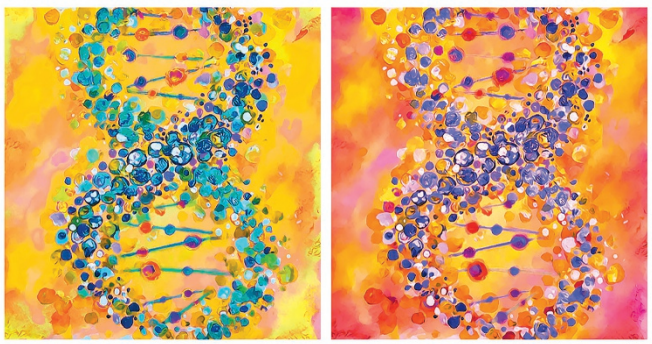
Despite rapid advances, not all genetic diseases can be detected using next-generation sequencing (NGS). Alterations in genes with highly repetitive sequences or in two nearly identical genes can make getting an accurate result challenging in a clinical setting. Such is the case with spinal muscular atrophy (SMA), a relatively common disorder with an incidence of about 1 in 10,000 and a carrier frequency varying from 1/40 to 1/100 in different ethnic groups. Two nearly identical genes, *SMN1* and *SMN2*, can contribute to disease incidence and severity. Current diagnostic methods include polymerase chain reaction (PCR) coupled with restriction fragment-length polymorphism analysis. Due to difficulty using NGS sequencing, SMA carrier detection has not been included in NGS panels to date. Here, Feng *et al.* report the development of an NGS-based carrier screen with nearly 100% sensitivity and specificity. The approach, they note, is amenable to being integrated into existing NGS-based carrier screening panels. The researchers validated the new method, called paralogous gene copy-number analysis by ratio and sum, in a clinical setting using 6,738 samples that included a cross-section of ethnic groups. Among these samples, African Americans and Hispanics had the lowest frequencies of SMN deletion, at 1.0 and 0.9%, respectively, whereas Asians had the highest, at 2.4%. The findings represent the first report of an NGS-based clinical method for copy-number analysis of genes with a high degree of sequence identity. *—Karyn Hede, News Editor*



NEWS BRIEFS

Do consumers give up rights to their genetic information by ordering ancestry testing?

Questions about consumer genetic privacy have generally been reserved for discussion within academic and policy circles. But the question of who owns DNA samples submitted for ancestry testing spilled over into social media recently, to the point that it rated an entry on Snopes, the fact-checking website. Earlier this year, an inflammatory blog post by attorney Joel Winston set off alarm bells among consumer protection advocates. He argued that submitting DNA to Ancestry.com amounted to forever surrendering rights to personal genetic information. The post, widely shared online and commented on in media circles, eventually prompted the company to change some of the language in its terms and conditions, including removal of language granting the company royalty-free rights in perpetuity. But many of the concerns raised in the original essay remain. The company's $99 test kit comes with a laundry list of items to which consumers must review before activating the kit. Those items include several layers of informed consent and checkboxes giving license for one's DNA to be included in research studies. Ancestry.com is hardly alone among direct-to-consumer DNA testing companies in its licensing terms. Whether consumers carefully read these terms before checking boxes remains an open question. Winston raises issues surrounding how consumers might revoke consent and whether, once genomic information has been stored and processed, revocation of rights may be meaningless. The company says that once genetic information is included in aggregated data, it cannot be removed. Further, the company acknowledges the "potential risk that third parties could identify you from research that is made publicly available." One thing is certain: in the words of Ancestry.com's Chief Privacy Officer Eric Heath, "There are a lot of legitimate questions to be asked and discussions to be had about consumer genomics." *—Karyn Hede, News Editor*

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