Expanded Carrier Screening - Optimizing Panel Design

James Goldberg, USA

Pan-ethnic expanded carrier screening is increasing being offered to maximize the identification of carrier couples who are at risk for having an affected offspring. Next generation sequencing (NGS) is commonly used to identify disease causing mutations in carrier couples. Important considerations in using NGS are detection of copy number variants and curation of identified variants. These issues will be discussed in relationship to designing a panel of disorders to test for. In addition, the overall advantages and limitations in using a NGS approach for carrier detection will be presented.