Abstract
Clinical genome and exome sequencing can diagnose pediatric patients with complex conditions that often require follow-up care with multiple specialties. The American Academy of Pediatrics emphasizes the role of the medical home and the primary care pediatrician in coordinating care for patients who need multidisciplinary support. In addition, the electronic health record (EHR) with embedded clinical decision support is recognized as an important component in providing care in this setting. We interviewed 6 clinicians to assess their experience caring for patients with complex and rare genetic findings and hear their opinions about how the EHR currently supports this role. Using these results, we designed a candidate EHR clinical decision support application mockup and conducted formative exploratory user testing with 26 pediatric primary care providers to capture opinions on its utility in practice with respect to a specific clinical scenario. Our results indicate agreement that the functionality represented by the mock-up would effectively assist with care and warrants further development.  

References