Continued coverage and reimbursement challenges for Diagnostic Exome Sequencing after 5 years, genetic counselors can help to overturn denied cases

Zöe Powis, Kelly Hagman, Taylor Cain, Layla Shahmirzadi, Sha Tang

Introduction: Since 2011, Diagnostic Exome Sequencing (DES) has proven beneficial in providing previously undiagnosed patients with molecular genetic diagnoses. Consequently, ACMG recommended proper utilization of DES in clinical assessment of individuals with suspected genetic conditions, where prior genetic testing failed to lead to diagnosis. Application of DES has allowed many undiagnosed patients who endured extensive genetic testing to receive definitive genetic diagnoses. Despite a belief of DES as cost-effective by the medical community, its utility, scientific and medical value is still questioned by payers.

Methods: 1500 consecutive patients that had undergone DES were retrospectively analyzed to determine payer category, excluding 24 research study exomes. Additionally, 538 cases were prospectively analyzed for denial, approval and appeal information.

Results: 911/1476 (61.7%) were approved by private insurance/Medicare/Medicaid, 521 (35.3%) institution billed, 44 (3.0%) paid directly by the patient/patient representation.

275/ 538 (51.1%) cases were approved for testing by private insurance/Medicare/Medicaid. Out of 267 denied cases, 65 cases appealed and 21 (32.3%) of these were subsequently approved.

Reasons for denial (several cases had multiple reasons) included: 176 (46.3%) experimental, 85 (22.4%) not medically necessary, 83 (21.8%) insurance policy exclusion/did not meet medical policy, 27 (7.1%) will not impact treatment, 5 (1.3%) other (e.g. no documentation given), 4 (1.1%) does not diagnose disease.

Discussion: Despite recommendations and literature demonstrating clinical utility of DES, out of the 48% denied cases, over 97% of those were denied due to insurance companies' rationalization that the DES does not have enough evidence to benefit patients. The number of overturned appeals is encouraging and eludes that genetic counselors can help in insurance authorizations and obtaining coverage for DES. While a number of insurances (both private and public) are covering DES, genetic counselors are needed to aid in education of payers and demonstration of the clinical utility of DES.