Use Case Workgroup Update

Marc S. Williams, MD
Chair
Current Use Cases under consideration or development

- Lynch syndrome
- Hearing loss and aminoglycoside exposure
- Smoking Cessation
- PKU
- Dental (with American Dental Association)
Lynch syndrome

• Lead: Nonniekaye Shelburne NCI
• Use case has gone through several cycles of review
• Current revision will be forwarded to ASCO representative (Greg Nowakowski) for review and consideration of sponsorship by ASCO
Hearing Loss

• Lead: Bracie Watson NIDCD
• Use case has gone through one set of revisions
• Seeking professional society sponsorship (American Academy of Pediatrics; American Academy of Otolaryngology-Head and Neck Surgery; Association for Research in Otolaryngology)
Smoking Cessation

• Lead: Joni Rutter NIDA
• Small group working on case refinement. Not presented to workgroup yet
• No professional society identified as yet
PKU

• Lead: Melissa Parisi NICHD
• Initial e-mails
• Use case development starting in May
• Potential Professional Society sponsorship (AAP; ACMG)
Dental

- Lead: Tom Hart American Dental Association; Emily Harris NIDCR
- ADA has educational cases in genetics that they would like to consider adapting into use cases
- Will review cases with Dr. Hart and choose one or more for use case development
Additional Informational Items

- Additional use case development partners
  - Illumina
    - Frank Ong lead
    - Attending Workgroup calls
    - Have some educational cases
      - Will review to identify ones that may work well as a use case
  - American Academy of Ophthalmology
    - Looking to develop use cases on rare genetic eye diseases (Retinoblastoma, Lebers Congenital Amaurosis, Juvenile X-linked Retinoschisis, Norries Disease, various Autosomal Dominant Diseases)
    - Paul Yang taking the lead
  - Cardiology (AJ Marian)
    - Extend invitation to join Use Case workgroup
  - AMP
    - 2 use cases submitted for review. Going through formal AMP review
  - Contacted Program Directors of IM. Not interested in participating formally