

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