

Matchmaker Exchange Tiered Informed Consent Proposal

The API WG has put forth the following proposal for a tiered approach to consenting patients for genomic matchmaking. The need for explicit patient consent will be dependent on the degree of potentially identifiable information shared with the matchmaking entity.

The first step of matchmaking involves a search being conducted by a data requester to establish the existence of similar patients in a collection of patient records. Once a discovery hit has occurred, it is typically then desirable for actual patient data to be exchanged between the data depositor and the data requester.

- Level 1 - Undertaking matchmaking based on top level HPO terms and/or candidate gene names - consent not required
 - Broad phenotype description - as a disease name (OMIM or Orphanet e.g. Charcot-Marie-Tooth disease) or by using top level HPO terms (axonal motor neuropathy)
 - HGNC approved gene names - for the suspected or candidate pathogenic loci
 - This level of information is essentially non-identifiable and therefore its use in data discovery (or even data sharing) implies no significant risk of harm and so such uses should not require explicit patient consent
 - Subsequent exchange of this level of information between depositor and requester (i.e., data sharing) raises no additional consent risks
 - Subsequent exchange of more detailed information between depositor and requester (i.e., data sharing) may require consent (see below)
- Level 2 - Undertaking matchmaking based on any depth of HPO terms and/or DNA or protein sequence level information including genomic variant datasets - consent required
 - Detailed phenotype description - using any set of HPO terms
 - Genomic variant dataset - including one or more variants (irrespective of suspected etiologic role), or related information such as variant class, amino-acid alteration, variant location, affected exon, etc.
 - This level of information is identifiable and therefore its use in data discovery implies a possible risk of harm and so such uses would require explicit patient consent
 - Subsequent exchange of this level of information between depositor and requester (i.e., data sharing) likewise would require consent
 - However, consent for matchmaker discovery and subsequent sharing of level 2 data can be taken to have been given if consent exists for the data to be included in an open or controlled access database whose declared purpose involves data sharing for purposes consistent with matchmaking