PROPOSED USE CASE TEMPLATE AND EXAMPLE

INTER-SOCIETY COORDINATING COMMITTEE, USE CASE WORKGROUP
(12/18/2013)

Template

I. Specialty/Professional Society

II. Type of Use Case
   a. Genomic-based therapeutics.
      i. Pharmacogenomic
   b. Rare Single Gene Mendelian Disorder
   c. Family History
   d. Common Complex Disease with Genetic Component
   e. Whole exome/genome sequencing
      i. Incidental findings (specifically included in EPA for genomic testing
   f. Microbial Genomics

III. Title

IV. Clinical Scenario

V. Description of relevant genomic information and how this information would be used

VI. Recommended clinical action
   a. Identification of patients within the clinician’s practice for which use case is relevant
   b. Specialty that develops use case could also define practice-based learning objectives that could be implemented based on the use case (e.g. Maintenance of Certification)

VII. Family Implications

VIII. Evidence to support the use of genomic information in this scenario
   a. Professional society practice guideline
   b. Other guideline
   c. Evidence review
   d. Consensus Best Practice

IX. List of additional resources
Example

I. **Specialty/Professional Society**: American Academy of Pediatrics

II. **Type of Use Case**: Pharmacogenetics

III. **Title**: HLA-B*1502 and adverse events related to use of Carbamazepine

IV. **Clinical Scenario**: A 16 year old Asian boy presents to the office with new onset of psychomotor seizures. He also has a history of a mood disorder. He is currently on no medications and has no known allergies or other contraindications. Based on this information the decision was made to initiate treatment with Carbamazepine.

V. **Relevant Genomic Information**: Severe, life-threatening skin reactions, including fatal cases, have occurred in patients treated with carbamazepine. These have included cases of Stevens-Johnson syndrome and toxic epidermal necrolysis. In countries with primarily Caucasian population the incidence is 1-6 per 10,000 new users. The rate approaches 10 times higher in Asian and Asian Indian populations. Patients who carry the HLA-B*1502 allele and are of Asian ethnicity appear to be at high risk for developing severe hypersensitivity skin reactions to carbamazepine. Patients of Asian ethnicity should be screened for this HLA type prior to initiation of carbamazepine. FDA has revised the boxed warning to reflect this risk.

VI. **Recommended Clinical Action**: Screening for the HLA-B*1502 allele should be performed in patients of Asian ethnicity prior to starting carbamazepine. If this is present an alternative medication is indicated unless the benefits of carbamazepine clearly outweigh the risks.

VII. **Family Implications**: None

VIII. **Supporting Evidence**: Guideline from the Clinical Pharmacogenetics Implementation Consortium (reference below)

IX. **References and Resources**
   b. FDA Label information. [http://www.pharmgkb.org/label/PA166104780](http://www.pharmgkb.org/label/PA166104780)