Special topics workgroup goal
To develop a proposal regarding the level of granularity necessary in the System part of LOINC codes in different domains of genetic testing, including germline versus somatic, in order to clarify the usage of existing codes and develop guidelines for determining whether new codes are necessary going forward.

Stimulus for creating this workgroup
1) Questions from the LOINC community regarding the use of Bld/Tiss codes, especially in relation to Saliva, Buccal swabs, and Bone marrow specimens, and requests to update Bld or Tiss codes to Bld/Tiss.

2) Requests from the LOINC community to update Bld/Tiss codes to Bld/Tiss/Saliva.

Questions we addressed
1. What level of specificity is needed in the System for germline genetic testing, such as for HLA, cytochrome p450, and heritable conditions, when in theory, the result should be the same in virtually all the cells in the body?

2. If we use a single generic System for germline testing, should we continue to call it Bld/Tiss? Should we define it as encompassing any specimen?

3. What level of specificity is needed for somatic mutation testing, e.g., for cancer? Is it important to distinguish different types of cancers (e.g., breast versus colon)? What about blood versus bone marrow?
Existing Systems in LOINC for genetic testing concepts

Germline
- Bld (~75)
- Bld/Tissue (1500+)
- Bld/Tiss/Saliva (11)
- Amnio (17)
- Amnio/CVS (~30)

CVS (7)

Somatic
- Bld/Tissue (~30)
- Bone marrow (~50)
- Tissue (~20)
- XXX (~20)

Cancer.XXX (~300 for genetic testing; updating to Cancer specimen for 2.60 release)
- Breast cancer specimen (<5)
- Colorectal cancer specimen (5)

Process
1. Reviewed the definitions of germline and somatic genetic testing.
2. Looked at different sources, including the Genetic Testing Registry, large laboratories, and laboratories associated with large children’s hospitals, to get a sense of the level of granularity at which these Systems are usually distinguished.
3. Contacted experts in the field and, if they responded, reviewed their feedback.
4. Determined what we (the workgroup) thought was the best way forward.
Summary

Our discussion mostly focused on germline testing.

In the area of germline testing, the key points we considered were that in theory, the result should be the same regardless of the specimen tested (not taking the quality of the specimen into account), and that if the test is done on one specimen, e.g., blood, it would most likely not be repeated on another type of specimen.

Various members of the HL7 Clinical Genomics workgroup consistently felt that for germline testing, LOINC should not create codes with specific Systems.

General consensus that it would make sense to include “Cells” in the System name to clarify what we are already doing and recommending in practice.

We also discussed the movement towards using the SPM segment more consistently, and that once enough institutions are using the SPM segment, less specificity may be okay in the LOINC System.

Regarding somatic (cancer) testing, the general consensus was that the type of cancer is important for interpreting the test results and that we should continue to use more specific Systems in LOINC (e.g., breast cancer specimen, bone marrow, etc.). However, there is concern about using “cancer” in the System name if it’s possible that the test comes back negative (in which case it isn’t cancer), and about post-treatment testing.

Recommendations

1. Continue to use a generic System in LOINC for germline testing.

2. Change the System name to Bld/Tiss/Cells to clarify the meaning and align with the current use of the term.

3. Define Bld/Tiss/Cells as:

   The LOINC ‘Bld/Tiss/Cells’ System includes blood, all types of tissue, and cells contained in swabs as well as fluids such as saliva. This System is primarily used for germline genetic testing, in which, specimen quality aside, the result should have the same meaning.
regardless of the type of specimen used. In general, this System will not be used for somatic or cancer testing since knowing the specific specimen type (e.g. colorectal cancer specimen or bone marrow) is important for interpreting the significance of the test result.

4. Consider addressing Systems for somatic testing in the future.