Proposal for LOINC Lab Committee regarding
System for human
genetic testing in LOINC
June 8, 2017

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.