A Coding Solution for Genetic Testing

A coding engine for accurate and consistent coding of genetic tests

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CONCERT
GENETICS

EXECUTIVE SUMMARY

The genetic testing market is growing fast and evolving rapidly. With more than 150,000 clinically orderable genetic tests on the market,[1] stakeholders are struggling to communicate effectively and efficiently across the Genetic Health Information Network.[2] One significant challenge is coding genetic tests for reimbursement. This is seen in claims data, where variation in coding correlates with variation in pricing.[3]

Concert Genetics, a leader in data and digital infrastructure for the Genetic Health Information Network, has developed a novel method to translate a genetic test into a single code or code combination. This method, delivered as the Concert Coding Engine, operates according to concise guiding principles, relies on finite data elements, and generates repeatable output, all without requiring stakeholders to adopt an additional unique identifier. This paper describes the Coding Engine, including the objectives, guiding principles, data inputs, underlying logic, and the limitations of the current implementation. The paper concludes with a brief discussion on how stakeholders may access and deploy the Concert Coding Engine.

OBJECTIVES & GUIDING PRINCIPLES

The overarching goal of a data-driven coding solution for genetic testing is to advance precision medicine through transparency and efficiency. In more concrete terms, the desired outcome is to achieve:

1. Predictable reimbursement according to market-based pricing, reducing administrative and financial costs for laboratories and insurers
2. Consistent application of medical policy with minimal human intervention, limiting time consuming and often manual reviews (i.e. prior authorization)
3. Accurate determination of insurance coverage and price, minimizing surprise bills received by patients

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[1] Concert Genetics data. 150,182 GTUs as of June 2020. Concert uses the term Genetic Testing Unit (GTU) for any orderable combination of analytes (e.g., genes or other targets) and techniques at a specific point in time, sold by a laboratory as a single item in the laboratory's catalog.
[2] Concert Genetics. May 2017. Connecting the Genetic Health Information Network: Critical Steps to Realizing the Potential of Precision Medicine. Available at: https://www.concertgenetics.com/blog/connecting-genetic-health-information-network-whitepaper/
[3] Concert Genetics. September 2019. Coding Variability in Genetic Testing: A summary of coding and price variation in commercial health insurance claims.

The Concert Coding Engine standardizes the coding process for genetic testing, allowing stakeholders to arrive at a single way to code each test on the market. To support these ends transparently, Concert utilized the following principles to guide development of the Coding Engine:

1. There is a single, repeatable way to code each genetic test on the market.
2. The relevant unit for coding is the test performed. The test performed is the test for which a patient receives a result (not a subset or component of the test performed, e.g., a multi-gene panel test is one unit).
3. Test attributes (genes, techniques and, for panel tests, clinical application), not attributes like patients' medical or family history, are the basis for assigning codes.
4. The Current Procedural Terminology (CPT®)[4] Code Book and supporting guidance can (and must) be translated into data structures that can be matched with known test attributes.
5. Choose simplicity over complexity, whenever possible. Panel codes are used whenever applicable. Single gene codes will ONLY be used when a multi-gene test does not meet criteria for any panel code. A maximum number of codes will be applied.

CODING ENGINE

Overview

The Coding Engine is a software application that transforms data inputs (test attributes) into a specific code or set of codes. The Coding Engine consists of three core elements: a database of genetic tests (and test attributes), expert-curated coding guidance, and a logic-driven algorithm for mapping tests to codes. Each of these elements is described below.

Sources of Genetic Test Data

The Coding Engine utilizes Concert's database of genetic tests as its source of tests and test attributes. While any sufficiently detailed database could be substituted, Concert's database has three primary benefits. First, Concert maintains an accurate current and historic snapshot of the entire market with minimal administrative burden on the laboratory industry.[5] Second, Concert maintains a high level of detail – not only all targets and techniques, but more than 20 other data elements – on each test in the database. Third, Concert combines additional data sources (e.g., claims, orders) to validate accuracy and estimate the frequency of gene utilization across the market. Each of these elements contributes to the accuracy and reliability of the Coding Engine.

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[4]CPT copyright 2020 American Medical Association. All rights reserved. CPT is a registered trademark of the American Medical Association.
[5] Users are invited to review Concert's genetic test database at www.ConcertGenetics.com at no charge. Users may report any test data errors via the website or directly to Concert Genetics. Most tests are updated on a weekly basis.

Sources of Coding Guidance

The Coding Engine requires the following data assets, each of which must be assembled and synchronized with the CPT® Code Book:[6]

Table 1: Required Data Elements

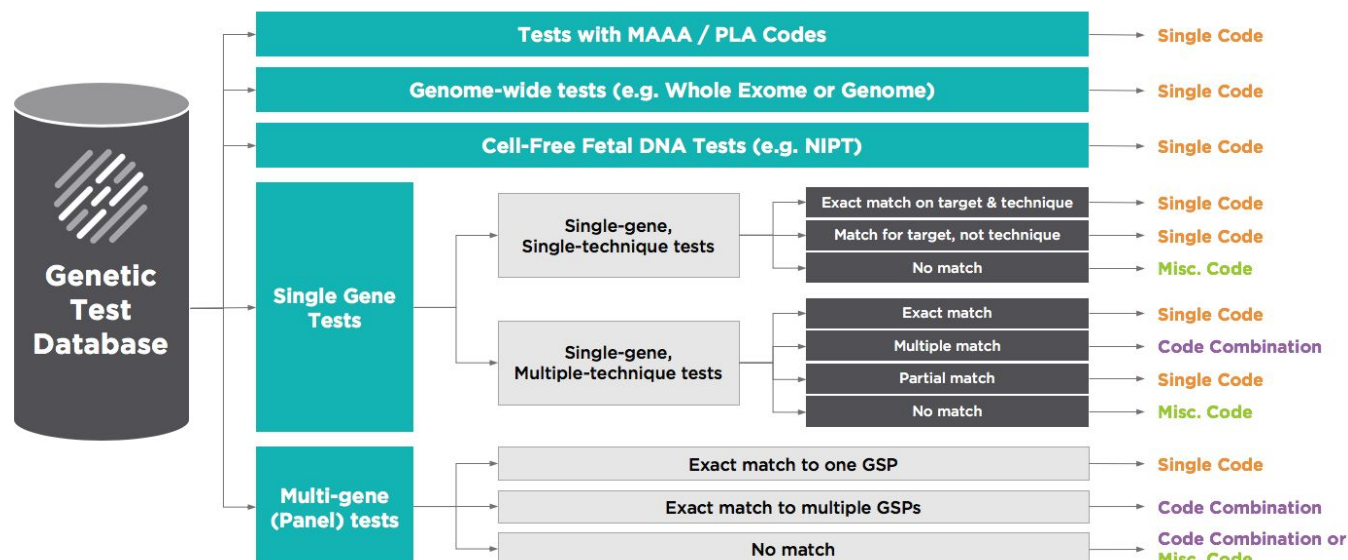| Data Table	Fields
PLA/MAAA Code Table	PLA or MAAA Code Lab Name Test Name
Tier 1 Code Table	Tier 1 Code Gene Name (HGNC Standard) Technique
Tier 2 Code Table	Tier 2 Code Gene Name Technique
Genomic and GSP Code Table	GSP Code Genes or Genomic Target Technique(s) Clinical Application
Gene Priority Score	All genes ordered by technique (sequencing, del/dup, targeted mutation) and by frequency of use (Concert database)

Coding Logic

The Coding Engine is driven by an algorithm, which is based on a logic that maps tests to codes. The logic is as follows:

1) <u>Tests with MAAA/PLA codes.</u> If the test has a specified MAAA or PLA code, assign that code.

2) <u>Genome-wide tests.</u> If the test covers the whole exome or genome, code as appropriate for the technique to match the exome, genome or microarray code(s).

3) <u>Cell-free fetal DNA tests.</u> If the test is designed to examine circulating cell-free fetal DNA (also known as non-invasive prenatal testing), it will be matched to the appropriate corresponding code or codes (aneuploidy and/or microdeletions).

4) <u>Single-gene tests.</u>
   a) <u>Single-gene, single-technique tests.</u> Assign the appropriate Tier 1 or Tier 2 code based upon the specific gene and technique provided.
      i) <u>Exact match on target and technique</u> – If there is an exact match, this code will be mapped.

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[6] The following code sections of the CPT® code book are relevant for the coding engine: Tier 1 Molecular Pathology , Tier 2 Molecular Pathology, Multi-Analyte Algorithmic Assays, Genomic Sequencing Procedures, Proprietary Laboratory Analysis Codes

    ii)   <u>Match for target, not technique</u> – If there is a target match, but a technique mismatch, a code may be used if the technique specified in the test supersedes the available code (e.g. If there is a targeted mutation code specific to a gene, but not a sequencing or del/dup code, the targeted mutation code can be used).

    iii)   <u>No match</u> – If there is no target or technique match, or if there is a target match, but a different technique in the specified test, use the miscellaneous code.

b)   <u>Single-gene, multiple-technique tests.</u> Assign the appropriate codes Tier 1 or Tier 2 codes, assigning multiple codes for distinct techniques performed. The broadest code (for which there is a code) will be coded. Sequencing codes and targeted mutation codes for the same gene will not be used in combination with one another.

    i)   <u>Exact match</u> – If there is only one code to cover sequencing and deletion / duplication, that code will be used before using individual single gene - single technique codes.

    ii)   <u>Multiple match</u> – If there are multiple codes for different techniques for the same target and those differing techniques are performed, those codes may be coded in conjunction with one another. Deletion/duplication analysis performed on NGS sequence data is considered a separate technique.

    iii)   <u>Partial match</u> – If there is a Tier 1 or Tier 2 code for one of the techniques included in the test, but not for the other, the test may be coded with one Tier 1 or Tier 2 code.

    iv)   <u>No match</u> – Use a single miscellaneous code to represent all components of the test.

5)   <u>Multi-gene (panel) tests.</u> Compute all target-technique combinations and match to similarly computed GSP target-technique combinations

a)   <u>Exact match to one GSP</u> – Assign GSP

b)   <u>Exact match to multiple GSPs</u> Two GSPs may be used if they represent different techniques for the same overlapping genes (e.g. 81435 and 81436). If these codes represent the same technique, the more specific of the indications for testing will be used (e.g. 81442 for Noonan Syndrome before 81443 for severe inherited conditions).

c)   <u>No match</u> – If there is no match, individual single gene codes may be used. All gene technique combinations will be made and mapped to the appropriate codes. Codes will be sorted by technique (Sequencing>Del/Dup> Exon>Targeted Mutation) and then by priority of gene frequency in the market. The first 5 matching single gene codes will be applied. If there are no appropriate single gene codes, a single miscellaneous code is used to represent all components of the test.

Figure 1: Logic Flow

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Genetic Test Database →

- Tests with MAAA / PLA Codes → Single Code
- Genome-wide tests (e.g. Whole Exome or Genome) → Single Code
- Cell-Free Fetal DNA Tests (e.g. NIPT) → Single Code
- Single Gene Tests
  - Single-gene, Single-technique tests
    - Exact match on target & technique → Single Code
    - Match for target, not technique → Single Code
    - No match → Misc. Code
  - Single-gene, Multiple-technique tests
    - Exact match → Single Code
    - Multiple match → Code Combination
    - Partial match → Single Code
    - No match → Misc. Code
- Multi-gene (Panel) tests
  - Exact match to one GSP → Single Code
  - Exact match to multiple GSPs → Code Combination
  - No match → Code Combination or Misc. Code

Summary Statistics and Limitations

The Coding Engine currently assigns discrete code combinations to 138,648 tests in Concert's database of 150,182 tests. The first public version of the Coding Engine is limited to molecular tests, although future versions may include remaining areas of testing such as cytogenetic tests (e.g. FISH) and immunohistochemistry. The Coding Engine assigned 754 unique code combinations for this universe of tests, contrasting starkly with more than 36,000 unique code combinations observed in a large database of genetic testing claims.[7]

USE OF THE CODING ENGINE

Concert aims to make the Coding Engine widely available, balanced with the need for a business model that allows for ongoing support of the application. As such, access to the Coding Engine is provided to laboratories, for use on a laboratory's own tests, without charge.[8] Health plans can license (for themselves or their delegates) the Coding Engine for an annual fee. Other parties with alternative applications (e.g., researchers) should contact Concert directly.

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[7] Concert Genetics. September 2019. Coding Variability in Genetic Testing: A summary of coding and price variation in commercial health insurance claims.
[8] Fees may apply for multiple users or for access to other Concert products & services.

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CONCLUSION

The Coding Engine is just one step toward further transparency in genetic testing and precision medicine. Challenges and shortcomings should be expected. With that in mind, Concert restates a commitment to working with all stakeholders to improve the Coding Engine, and, more importantly, to advance the digital infrastructure necessary for the success of precision medicine.