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Potential Updates to ICD-10 Codes for Cystic Fibrosis and Related Disorders

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Abstract

Cystic fibrosis is one of the most common genetic disorders. It usually affects a number of body systems. The current ICD-10 classification of cystic fibrosis is based on manifestations. However, an expert WHO report from June 2000 ("Classification of Cystic Fibrosis and Related Disorders") recommended changing the structure for classifying cystic fibrosis in a future revision of ICD, to identify atypical and classical cystic fibrosis, with the latter further identified as with or without pancreatic insufficiency. A number of other areas also need to be considered within the scope of classifying cystic fibrosis, to enable better handling of disorders related to cystic fibrosis, particularly cystic fibrosis transmembrane conductance regulator (CFTR) mutations.

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Introduction

Cystic fibrosis is recognized as one of the most common genetic disorders. It usually affects a number of body systems, with the pulmonary and gastrointestinal systems being just two that are commonly affected. Cystic fibrosis is classified in ICD-10 to category E84, Cystic fibrosis. The current structure of this category is based on manifestations. A number of other areas need to be considered within the scope of classifying cystic fibrosis, to enable better handling of disorders related to cystic fibrosis.

At this time, screening for cystic fibrosis is often performed on neonates. Thus, it may be found when it is asymptomatic. In classical cystic fibrosis, some pulmonary manifestations develop within the first year, with more significant manifestations developing subsequently, often after ten years or more. While some cases present in neonates with meconium ileus, it is also possible to have a similar ileus in older individuals. Gastrointestinal manifestations can be related to pancreatic insufficiency, which occurs in many cases, but is not universal. Some cases do not have the severe pulmonary manifestations of classical cystic fibrosis, and these cases may be referred to as atypical cystic fibrosis.

In a WHO report from June 2000 ("Classification of Cystic Fibrosis and Related Disorders"), experts recommended changing the structure for classifying cystic fibrosis in a future revision of ICD to identify classical cystic fibrosis with pancreatic insufficiency, classical cystic fibrosis without pancreatic insufficiency (pancreatic sufficient), and atypical cystic fibrosis (also with codes for other and unspecified cystic fibrosis). Also, a number of disorders were identified as related to the cystic fibrosis gene, that is to cystic fibrosis transmembrane conductance regulator (CFTR) mutations. While these disorders are not considered cystic fibrosis, the connection with the CFTR gene is of clinical importance.

This paper reviews potential ways that ICD-10 could be brought up to date with current clinical knowledge for cystic fibrosis and related disorders.

Issues for Consideration for ICD-10 Updating

Hypertrypsinaemia

In neonates and occasionally in older individuals, trypsin may be checked as part of screening for cystic fibrosis. Hypertrypsinaemia may indicate a need for further testing, and in some cases for which cystic fibrosis is not diagnosed, it may be the only thing reported. It will be beneficial to differentiate neonatal hypertrypsinaemia, since this has different implications in a neonate than in an older individual.

Proposed new codes for hypertrypsinaemia
E16 Other disorders of pancreatic internal secretion E16.5 Hypertrypsinaemia Excludes: neonatal hypertrypsinaemia (P96.6) P96 Other conditions originating in the perinatal period P96.6 Neonatal hypertrypsinaemia

Pancreatic Insufficiency

It would help to better track the effects of cystic fibrosis to create an asterisk code for secondary pancreatic insufficiency, to be used along with an appropriate E84 code to show it is related to cystic fibrosis. This could also be used with certain other disorders.

Proposed new asterisk code for pancreatic insufficiency

K87* Disorders of gallbladder, biliary tract and pancreas in diseases classified elsewhere

K87.2*, Secondary pancreatic insufficiency

Of the current E84 codes, the ones that would appear to be appropriate to use as dagger codes are E84.1, Cystic fibrosis with intestinal manifestations, and E84.8, Cystic fibrosis with other manifestations. Additional proposed new codes will be considered subsequently.

Considerations for Future ICD Revision

Cystic Fibrosis (ICD-10 Category E84)

There are a few options for changing E84, none of which have had unanimous support from the experts consulted. A number of experts stated that the current codes are not useful clinically, due to the fact that most patients have manifestations involving multiple systems. Thus if full detail is known, most cases should be classified to E84.8, Cystic fibrosis with other manifestations. Some experts would favor deactivating current codes at E84 (E84.0 and E84.1), and restructuring it to follow the approach described in the June 2000 WHO report (mentioned in the introduction above). That approach would eliminate the manifestations from the codes. However, there could be negative effects for some uses of the data, if current codes were deactivated. These options may need further consideration.

If a newborn (or other person) has no obvious clinical symptoms, but based on their lab results is diagnosed with cystic fibrosis, it could be helpful to differentiate the cystic fibrosis as being subclinical or asymptomatic.

The approach shown below avoids major changes to the current codes, but proposes new codes for specific terms that would currently be presumed to go to E84.8.

One option for potential new codes for category E84, Cystic fibrosis

E84 Cystic fibrosis

Includes: mucoviscidosis

E84.0 Cystic fibrosis with pulmonary manifestations

E84.1 Cystic fibrosis with intestinal manifestations

Meconium ileus† (P75*)

Excludes: meconium obstruction in cases where cystic fibrosis is known not to be present (P76.0)

E84.2 Classical cystic fibrosis with other manifestations

Classical cystic fibrosis with multiple manifestations

E84.3 Atypical cystic fibrosis with other manifestations

Atypical cystic fibrosis with multiple manifestations

E84.4 Subclinical cystic fibrosis

Asymptomatic cystic fibrosis

Cystic fibrosis with no clinical manifestations

E84.8 Cystic fibrosis with other manifestations

Cystic fibrosis with combined manifestations

E84.9 Cystic fibrosis, unspecified

Another issue of potential concern is what clinical terms are likely to be used in medical records for these forms of cystic fibrosis. It is not clear how often clinicians will document using terms such as classical, atypical, and subclinical cystic fibrosis. Records frequently may not give detail, leading to the common use of code E84.9, Cystic fibrosis, unspecified.

The issues described above need further consideration in determining the best approach for updating the cystic fibrosis category.

For the proposed new K87.2*, Pancreatic insufficiency related to cystic

fibrosis, it is also important to consider which of these new proposed E84 codes would be appropriate to use as dagger codes along with the new K87.2*. It would appear to be appropriate to use the potential new code E84.2, Classical cystic fibrosis with other manifestations, and code E84.3, Atypical cystic fibrosis with other manifestations, as a dagger codes with the proposed new code K87.2*.

CFTR mutations

Related to coding for CFTR mutations, some of these may be coded using the proposed hypertrypsinaemia codes. One option that could be considered more broadly for genetic diseases would be to create a new section related to genetics in the U code section, and a new specific code for CFTR mutation (and cystic fibrosis carrier). Related to genetics, it would only be possible to create specific codes for a small number of the more common genetic disorders. There would be various ways this could be approached, and further expert input would be necessary to determine what code ranges should be used.

Option one for CFTR mutations and genetics
Provisional assignment of new diseases of uncertain etiology (U00–U40) Provisional assignment of new diseases of certain etiologies (U41–U49) Genetic susceptibility to disease and genetic carrier states (U41-U45) Excludes: sickle-cell trait (D57) U41 Cystic fibrosis transmembrane conductance regulator (CFTR) mutation Cystic fibrosis carrier

Another option that would not require such broad changes would be to create a new category at E91. Categories E91-E99 are unused. Section E70-E90 is Metabolic Disorders, within chapter 4, Endocrine, nutritional and metabolic diseases (E00–E90). This would not address genetic issues more broadly, but would involve less extensive changes.

Option two for CFTR mutations
Chapter 4 Endocrine, nutritional and metabolic diseases (E00–E91) … Metabolic Disorders (E70-E91) … E91 Cystic fibrosis transmembrane conductance regulator (CFTR) mutation Cystic fibrosis carrier

Further development of this approach could require excludes notes between the proposed hypertrypsinaemia codes, the CFTR code, and the cystic fibrosis codes, to make it clear that the more specific code should be used.

Future steps

At this time, it appears reasonable to consider adding new codes for hypertrypsinaemia. Additionally, it appears reasonable to consider adding a new code for secondary pancreatic insufficiency, to be used with certain of the existing cystic fibrosis codes (among potential others). These proposals could be submitted for URC consideration for the 2006 cycle.

At this time, there is not expert consensus on potential ways to update the classification of cystic fibrosis in ICD-10 (at category E84). While there was agreement that the current structure based on manifestations was not ideal, and a number of ways to change this were contemplated, there were

potential issues with proposed changes. Some of the issues raised, such as coding for subclinical cystic fibrosis, will need future consideration. It was not clear whether the desired improvement to the classification of cystic fibrosis could be handled within the constraints of a simple update to ICD-10. There was not clear consensus that the final recommendations from the 2000 approach would be optimal for a future revision of ICD, although that should be the starting point for further work.

Regarding disorders associated with CFTR mutations, these could be considered either in the context of a broader approach to genetic disorders, or just as a single issue to be added to the metabolic disorders. Wider input will be needed to determine the optimal approach.

Experts on cystic fibrosis and genetics consulted by conference call and e-mail include Victor Boulyjenkov (WHO), John Dodge (UK – University of Wales Swansea), Richard Olney (US – CDC), Beryl Rosenstein (US – John Hopkins University), Elizabeth Tullis (Canada – University of Toronto / St. Michael's Hospital), and Frank Accurso (US – University of Colorado / Children's Hospital of Denver). Classification experts consulted include Kerry Innes (Australia) and Lori Moskal (Canada). An additional stakeholder consulted is Suzanne Pattee (US – Cystic Fibrosis Foundation).

Revision Trial of Specific Clinical Topics

Cystic Fibrosis, in the Template for Reporting to URC

Preliminary

Identify Clinical Topic and Scope: chapter, block of codes, single code

Topic: cystic fibrosis and related disorders

Scope: Category E84, Cystic fibrosis, could potentially have codes added or modified.

A number of other areas could have codes added, to enable better handling of disorders related to cystic fibrosis.

Identification of Key Issues with current ICD Taxonomy: structure, terms, definitions, index, linkage, coding rules

The current structure of the cystic fibrosis category in ICD-10 is based on manifestations. In a WHO report from June 2000 ("Classification of Cystic Fibrosis and Related Disorders"), experts recommended changing this in future to identify pancreatic-insufficient classical cystic fibrosis, pancreatic-sufficient classical cystic fibrosis, and atypical cystic fibrosis (also with codes for other and unspecified cystic fibrosis). Also, there is a need to be able to represent disorders related to the cystic fibrosis gene, but which are not considered cystic fibrosis.

Findings as they relate to examination of other specialty modifications (ICD-O-3), national modifications (ICD-10-AM), other international classifications (ICPC-2)

We are not aware of any modifications of related specialty classifications for cystic fibrosis.

Identify any linkages between the clinical topic and the Map of

Medicine and SNOMED-CT: equivalency of terms, concepts, rules

SNOMED CT has some concepts similar to the current ICD-10 codes, but for the most part does not currently specifically represent the recommended new concepts. The Map of Medicine does not appear to contribute to these specific cystic fibrosis concepts, either.

Consultation Process: identify experts, groups, stakeholders, end-users and the role they played.

Experts on cystic fibrosis and genetics consulted by conference call and e-mail include Victor Boulyjenkov (WHO), John Dodge (UK – University of Wales Swansea), Richard Olney (US – CDC), Beryl Rosenstein (US – John Hopkins University), Elizabeth Tullis (Canada – University of Toronto / St. Michael's Hospital), and Frank Accurso (US – University of Colorado / Children's Hospital of Denver). Classification experts consulted include Kerry Innes (Australia) and Lori Moskal (Canada). An additional stakeholder consulted is Suzanne Pattee (US – Cystic Fibrosis Foundation).

List of Recommendations:

Proposed new codes for hypertrypsinaemia and neonatal hypertrypsinaemia.

E16 Other disorders of pancreatic internal secretion

E16.5 Hypertrypsinaemia

P96 Other conditions originating in the perinatal period

P96.6 Neonatal hypertrypsinaemia

Proposed new code for pancreatic insufficiency related to cystic fibrosis:

K87* Disorders of gallbladder, biliary tract and pancreas in diseases classified elsewhere

K87.2*, Pancreatic insufficiency related to cystic fibrosis

A final recommendation for approach to updating category E84 may require further development. One option for category E84 follows, but there was not a consensus agreement of the experts consulted.

E84 Cystic fibrosis

Includes: mucoviscidosis

E84.0 Cystic fibrosis with pulmonary manifestations

E84.1 Cystic fibrosis with intestinal manifestations

Meconium ileus† (P75*)

Excludes: meconium obstruction in cases where cystic fibrosis is known not to be present (P76.0)

E84.2 Classical cystic fibrosis with other manifestations

Classical cystic fibrosis with multiple manifestations

E84.3 Atypical cystic fibrosis with other manifestations

Atypical cystic fibrosis with multiple manifestations

E84.4 Subclinical cystic fibrosis

Asymptomatic cystic fibrosis

Cystic fibrosis with no clinical manifestations

E84.8 Cystic fibrosis with other manifestations

Cystic fibrosis with combined manifestations

E84.9 Cystic fibrosis, unspecified

List of Recommendations (continued)**:**

Handling of disorders related to the cystic fibrosis gene but which are not considered cystic fibrosis has more than one option possible, depending on whether a broader approach to genetics is attempted at the same time. One option could be to create a new section at the U codes, with a new category, as shown below.

Option 1

Provisional assignment of new diseases of uncertain etiology (U00–U40)

Provisional assignment of new diseases of certain etiologies (U41–U49)

Genetic susceptibility to disease and genetic carrier states (U41-U45)

Excludes: sickle-cell trait (D57)

U41 Cystic fibrosis transmembrane conductance regulator (CFTR) mutation

Cystic fibrosis carrier

Another option would be to create a new category at E91, to be part of the Metabolic Disorders, as shown below.

Option 2

Chapter 4

Endocrine, nutritional and metabolic diseases (E00–E91)

…

Metabolic Disorders (E70-E91)

…

E91 Cystic fibrosis transmembrane conductance regulator (CFTR) mutation

Cystic fibrosis carrier

Identify impact of recommended changes on the taxonomy of any other chapter, category, block, code, and rule within ICD-10.

The changes to the U codes noted above would be significant. Changes to other areas may be contemplated, such as adding asterisk codes, or indexing for certain conditions related to cystic fibrosis.

Identify a plan for field-testing the recommendations:

For future consideration.

Resource Requirements: professional fees, travel, teleconferences, meetings, man-hours

For future consideration.

List of Appendices (detailed documentation)

Under development.