

# 疾患・表現型データベース (Gendoo, OMIM, 他)

仲里 猛留

NAKAZATO, Takeru

@chalkless



情報・システム研究機構 データサイエンス共同利用基盤施設  
ライフサイエンス統合データベースセンター

Database Center for Life Science (DBCLS),  
Joint Support-Center for Data Science Research, Research Organization of Information and Systems (ROIS)

## イベント

統合データベース講習会：  
AJACS

受入れ機関募集

学会・展示会

トーゴの日シンポジウム

BioHackathon

NCIS 2016 オンライン講習会

その他のイベント

関連情報

＞ [講師派遣](#)

## 統合データベース講習会：AJACS徳島



<https://events.biosciencedbc.jp/training/ajacs76>

資料：<https://github.com/AJACS-training/AJACS76>

統合データベース講習会：AJACSは、生命科学系のデータベースやツールの使い方、データベースを統合する活動を紹介する講習会です。

今回の講習会では、生命科学系データベースのカatalog、横断検索、アーカイブ、ヒトデータに関するサービス・ツール等の紹介に加えて、多型データベース、疾患・表現型データベース、相互作用・パスウェイデータベース、ゲノムブラウザ・発現・局在関連データベース、タンパク質立体構造データベース、化合物データベースについてご紹介します。参加者全員がハンズオンでコンピュータを使いながらの講習です。

このセクションはtweet OK

ハッシュタグは #AJACS

# データベース統合化における問題点

- ・ 似たようなデータベースがたくさんありすぎる  
→ 何を見ていいのかわからない  
えてして全部見ないといけない…
- ・ そもそもどう見たらいいかわからない
- ・ データベース間でエントリ間の対応がとれない
- ・ 記述内容がバラバラで同じデータベース内でも対応がとれない
- ・ 文章（自然言語）で書かれていてコンピューターで一括処理できない

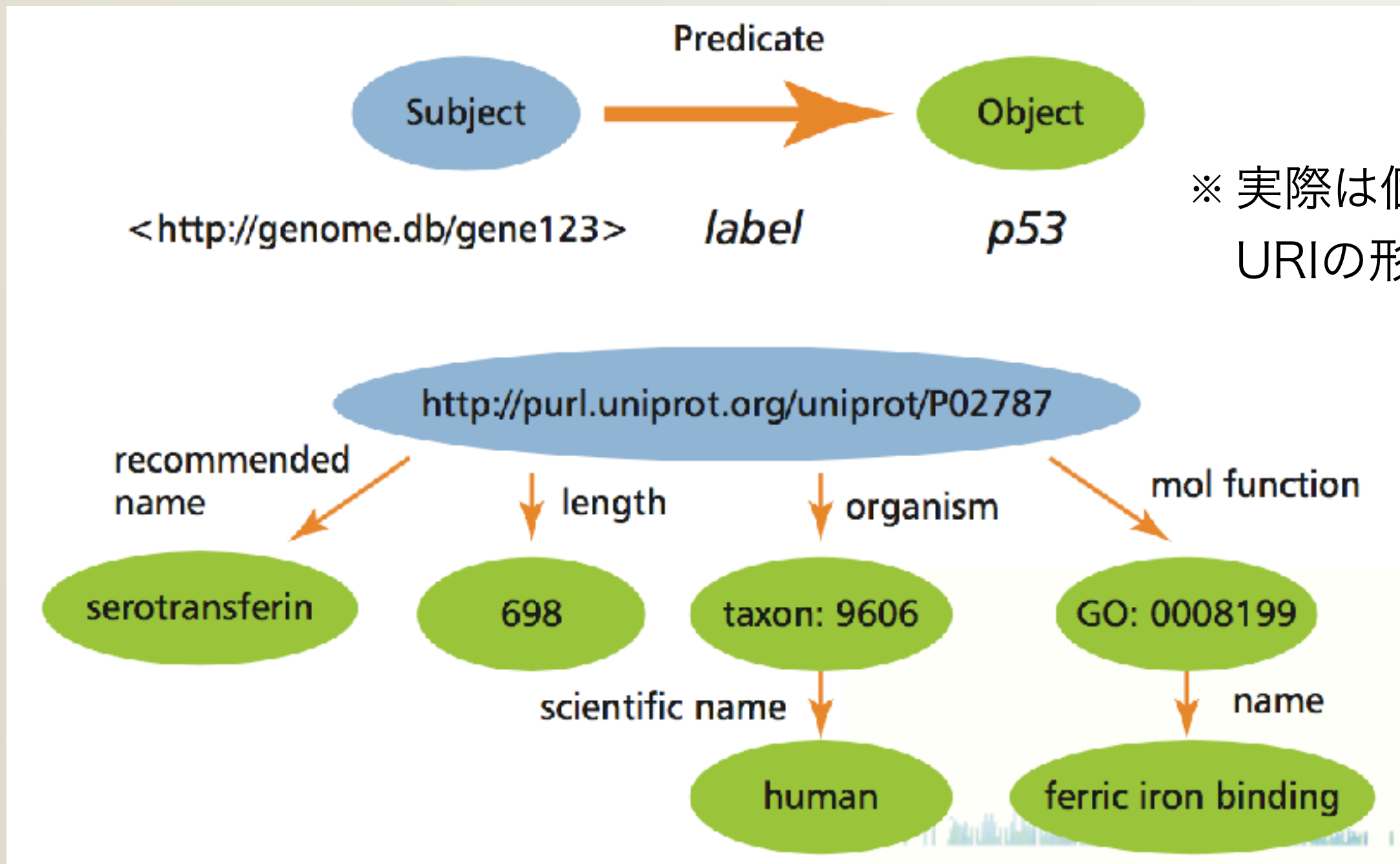
# 統合化を支えるSemantic Web技術

## RDF : Resource Description Framework

主語

述語

目的語



そんなこと  
言われましても



# 誰でもできるデータベース統合の考え方

別に Excel でデータを管理してもいい  
(あとで関係性をバラしてRDFにできる)

が、

同じことが書かれる列は同じラベルをつけておく  
(そしてできれば誰かのマネをする)

データ部分は既存の用語集から用語をもってくる  
せめて同じデータ内容は同じ記述をする

※ 用語集＝オントロジー

これだけで統合化がだいぶ楽に

# さまざまな疾患データベース

ICD-10 Version:2016

Search  (Advanced Search)

ICD-10 Versions - Languages Info

ICD-10 Version:2016

- I Certain infectious and parasitic diseases
- II Neoplasms
- III Diseases of the blood and blood-forming organs and certain disorders involving the immune mechanism
- IV Endocrine, nutritional and metabolic diseases
  - E00-E07 Disorders of thyroid gland
    - E00 Congenital iodine-deficiency syndrome
    - E01 Iodine-deficiency-related thyroid disorders and allied conditions
    - E02 Subclinical iodine-deficiency hypothyroidism
    - E03 Other hypothyroidism
    - E04 Other nontoxic goitre
    - E05 Thyrotoxicosis [hyperthyroidism]
    - E06 Thyroiditis
    - E07 Other disorders of thyroid
  - E10-E14 Diabetes mellitus
  - E15-E18 Other disorders of glucose regulation and pancreatic islet secretion
  - E20-E26 Disorders of other endocrine glands
  - E40-E46 Malnutrition
  - E50-E64 Other nutritional deficiencies
  - E65-E68 Obesity and other hyperalimentation
  - E70-E90 Metabolic disorders
- V Mental and behavioural disorders
- VI Diseases of the nervous system
- VII Diseases of the eye and adnexa
- VIII Diseases of the ear and mastoid process

E06.9 Thyroiditis, unspecified

E07 Other disorders of thyroid

E07.0 Hypersecretion of calcitonin

- C-cell hyperplasia of thyroid
- Hypersecretion of thyrocalcitonin

E07.1 Dysormogenic goitre

- Familial dysormogenic goitre
- Pendred syndrome

E07.2 Other specified disorders of thyroid

- Abnormality of thyroxine-binding globulin

About Statistics Downloads Contact Us MIMmatch Donate Help

Options Display ☐ Highlights

#274600

Table of Contents

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History

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Contributors

Create Date

Site History

# 274600

PENDRED SYNDROME; PDS

Alternative titles/synonyms

DEAFNESS WITH GOITER

GOITER-DEAFNESS SYNDROME

THYROID DYSFUNCTION

THYROID HORMONE DEFECT

HYPOTHYROIDISM

Phenotype-Gene Relationships

Location

Phenotype

Agg. 1

Pendred syndrome

Clinical Synopsis

TEXT

A number sign (#) is used hereafter to denote manual curation or computer generated text.

There is evidence that PDS is caused by a heterozygous mutation.

External Links

Protein

Clinical Resources

Clinical Trials

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Gene Reviews

Genetics Home Reference

OMIM

NCBI Resource How To

ModGen

ModGen

Search

Help

Full Report +

Pendred syndrome (PDS)

MedGen UID: 52850 • Concept ID: C0271529 • Disease or Syndrome

Synonyms:

Deafness with goiter; Oiler-deafness syndrome; HYPOTHYROIDISM, CONGENITAL, DUE TO DYSHORMONOGENESIS, 2B; PDS; Pendred Syndrome/DFNB4; Pendred's syndrome; SLC26A4-Related Pendred Syndrome; THYROID DYSHORMONOGENESIS 2B; THYROID HORMONE DEFECT, GENETIC DEFECT IN, 5B

Modes of inheritance:

Autosomal recessive inheritance (HPO: OMIM, Orphanet)

SNOMED CT:

Pendred's syndrome (70348004); Hypothyroidism with sensorineural deafness (70348004); Thyroid hormone organization defect II B (70348004); Oiler-deafness syndrome (70348004); Genetic defect in thyroid hormoneogenesis II B (70348004); DFNB4 (70348004); Pendred syndrome (70348004)

Genes (locations):

FOXI1 (5q35.1); KCNJ13 (1p23.2); SLC26A4 (7q22.3)

OMIM®:

274600

Definition

Pendred syndrome/horseshoe-shaped enlarged vestibular aqueduct (HSD/NEVA) comprise a phenotypic spectrum of sensorineural hearing loss (SNHL) that is usually congenital and often severe to profound (although mild-to-moderate progressive hearing impairment also occurs), vestibular dysfunction, and temporal bone anomalies (distal: enlarged vestibular aqueduct with or without cochlear hypoplasia). PDS also includes development of euthyroid goiter in late childhood to early adulthood whereas NEVA does not. [From GeneReviews]

Additional descriptions

From OMIM

Pendred syndrome, the most common syndromic form of deafness, is an autosomal recessive disorder associated with developmental abnormalities of the cochlea, sensorineural hearing loss, and diffuse thyroid enlargement (goiter) [Krivavt et al., 1997]. For a general phenotypic description and a discussion of genetic heterogeneity of thyroid dysmorphogenesis, see DFNB1 (274800). <http://www.omim.org/entry/274600>

From HPO

Pendred syndrome is a disorder typically associated with hearing loss and a thyroid condition called a goiter. A goiter is an enlargement of the thyroid gland, which is a butterfly-shaped organ at the base of the neck that produces hormones. If a goiter develops in a person with Pendred syndrome, it usually forms between late childhood and early adulthood. In most cases, this enlargement does not cause the thyroid to malfunction. In most people with Pendred syndrome, severe to profound hearing loss caused by changes in the inner ear (sensorineural hearing loss) is evident at birth. Less commonly, hearing loss does not develop until later in infancy or early childhood. Some affected individuals also have problems with balance caused by dysfunction of the vestibular system, which is the part of the inner ear that helps maintain the body's balance and coordinates eye movement.

Go to: [ ] [ ]

Go to: [ ] [ ]

Table of contents

Definition

Additional descriptions

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Term hierarchy

Professional guidelines

Recent clinical studies

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Genetic Testing Registry

Deletion/duplication analysis (19)

Microsatellite instability testing (MSI) (1)

Mutation scanning of select exons (1)

Research (1)

Sequence analysis of select exons (4)

Sequence analysis of the entire coding region (79)

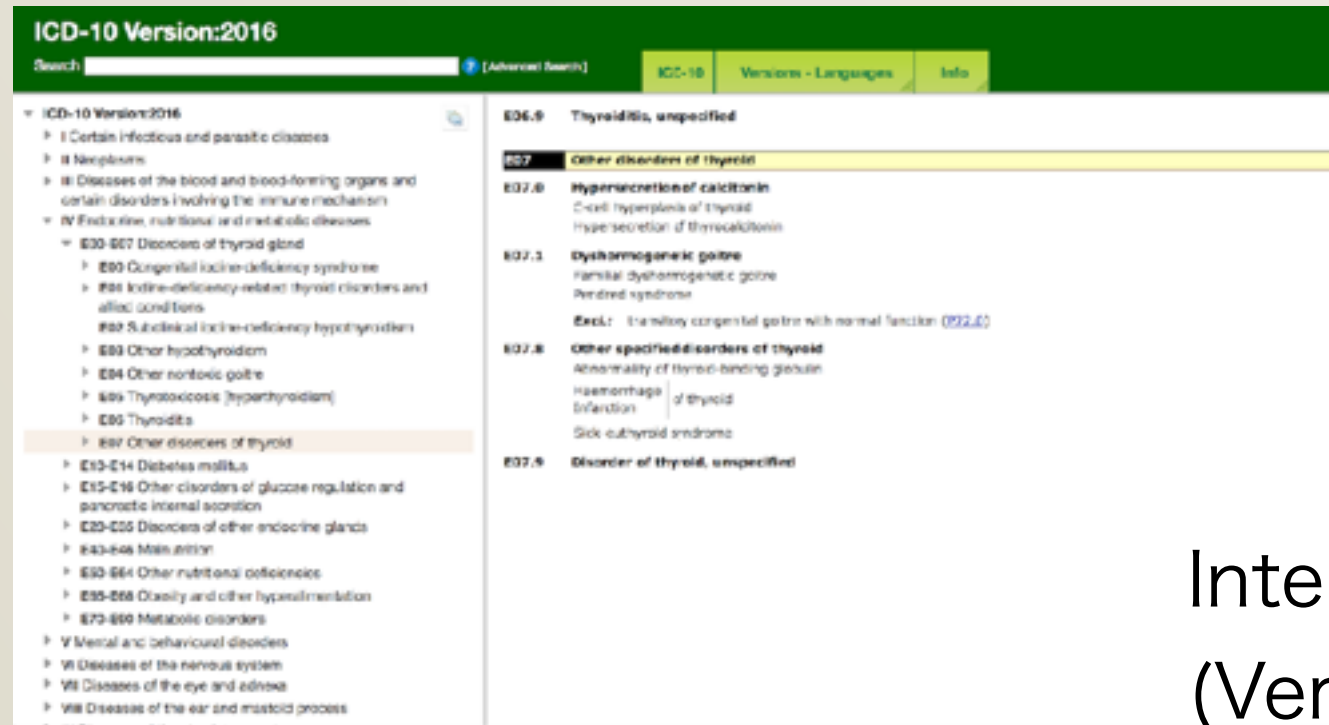
Targeted variant analysis (12)

See all (56)

Clinical resources



# ICD-10



## International Classification of Diseases (Ver.10)

<https://www.who.int/classifications/icd/icdonlineversions/en/>

by WHO

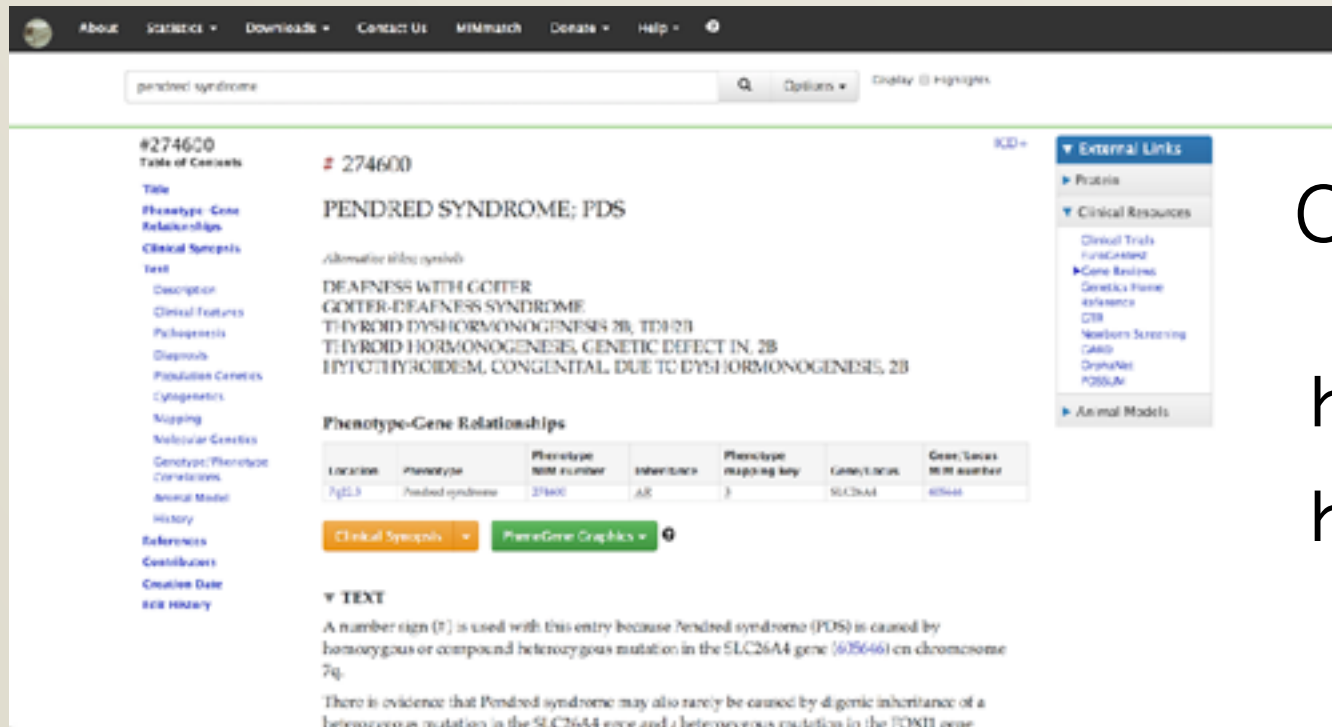
疾患名リスト

日本語化もされている

電子カルテ等でも病名の基準に

病名だけで、詳細な解説はない  
ましてや関連遺伝子はわからない

# OMIM



The screenshot shows the OMIM website interface. At the top is a navigation bar with links: About, Science, Downloads, Contact Us, MIMmatch, Donate, and Help. Below this is a search bar containing "pendred syndrome" and buttons for "Options" and "Display" (with a "Highlights" link). The main content area displays the entry for #274600, titled "PENDRED SYNDROME; PDS". It includes a "Table of Contents" on the left with links to Title, Phenotype-Gene Relationships, Clinical Synopsis, Text, Description, Clinical Features, Pathogenesis, Diagnosis, Population Genetics, Cytogenetics, Mapping, Molecular Genetics, Genotype/Phenotype Correlations, Animal Model, History, References, Contributors, Creation Date, and Edit History. The "Phenotype-Gene Relationships" section contains a table with columns: Location, Phenotype, Phenotype MIM number, Inheritance, Phenotype mapping key, Gene/Locus, and Gene/Source MIM number. The table lists one entry: 7q31.3, Pendred syndrome, 274600, AR, 3, SLC26A4, 605446. Below the table are buttons for "Clinical Synopsis" and "PhenGene Graphics". The "Text" section begins with a paragraph explaining the use of the number sign (#) and the cause of the syndrome by mutations in the SLC26A4 gene.

Location	Phenotype	Phenotype MIM number	Inheritance	Phenotype mapping key	Gene/Locus	Gene/Source MIM number
7q31.3	Pendred syndrome	274600	AR	3	SLC26A4	605446

Online Mendelian Inheritance in Man

<https://www.omim.org/>

<https://www.ncbi.nlm.nih.gov/omim/>

Dr McKusickらによる詳細な解説

あくまで遺伝子による疾患なので  
原因遺伝子により独自に  
ナンバリングされているものも  
例：NIDDM1/2/3

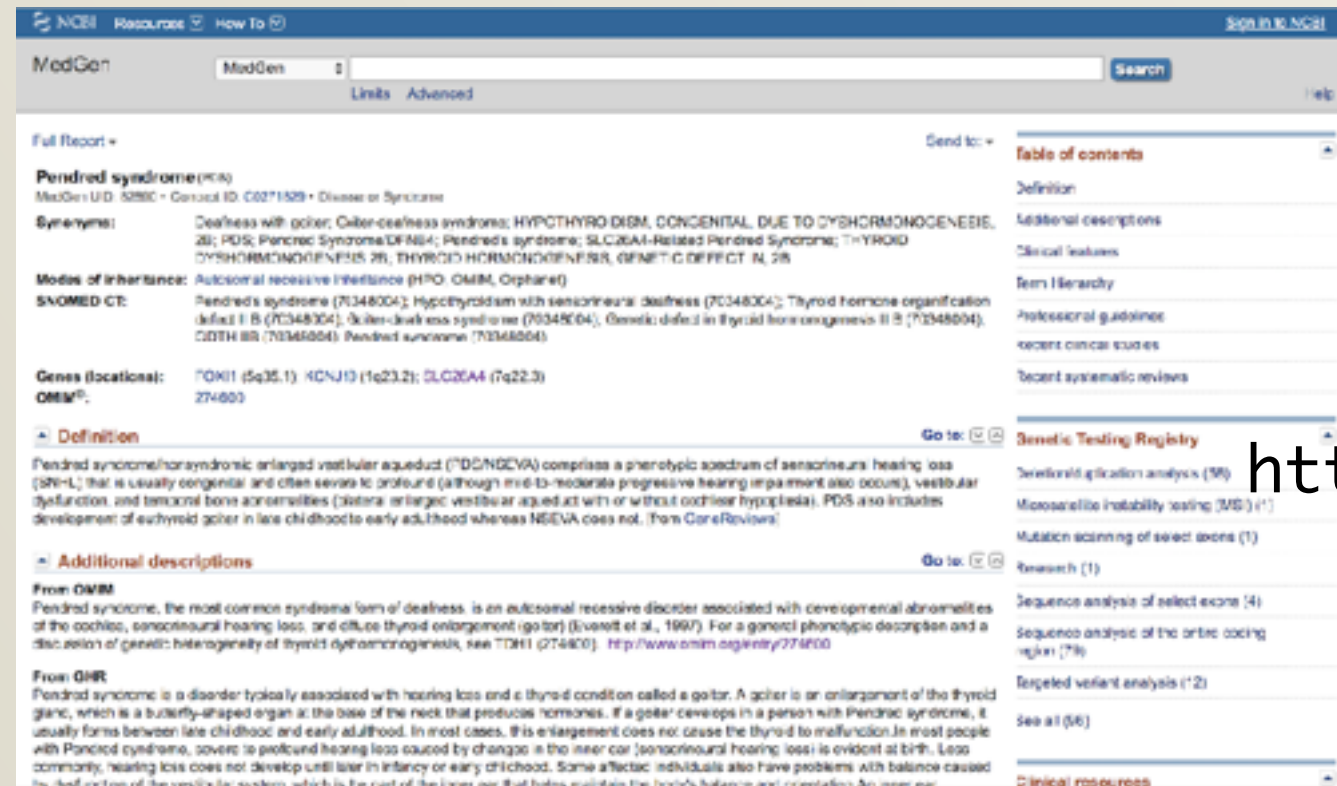
最近ライセンスがきびしくなった？

ヒト疾患と疾患関連遺伝子の  
データベース

Johns Hopkins 大学

26256 entries

# MedGen



The screenshot displays the MedGen interface for the entry "Pendred syndrome (PDS)". The top navigation bar includes "NCBI", "Resource", "How To", and a "Sign in to NCBI" link. The search bar contains "MedGen" and a "Search" button. The main content area is divided into several sections:

- Full Report:** A dropdown menu.
- Pendred syndrome (PDS):** MedGen UID: 52860, Concept ID: C0271529, Diseases or Syndromes.
- Synonyms:** Deafness with goiter; Oiler-deafness syndrome; HYPOTHYROIDISM, CONGENITAL, DUE TO DYSHORMONOGENESIS, 2B; PDS; Pendred Syndrome/DFNB4; Pendred's syndrome; SLC26A4-Related Pendred Syndrome; THYROID DYSHORMONOGENESIS 2B; THYROID HORMONOGENESIS, GENETIC DEFECT IN, 2B.
- Modes of inheritance:** Autosomal recessive inheritance (HPO: OMIM, Orphanet).
- SNOMED CT:** Pendred's syndrome (70348004); Hypothyroidism with sensorineural deafness (70348004); Thyroid hormone organization defect II B (70348004); Oiler-deafness syndrome (70348004); Genetic defect in thyroid homeogenesis II B (70348004); GDTH II B (70348004); Pendred syndrome (70348004).
- Genes (location):** PDK11 (5q35.1); KCNJ13 (16p23.2); SLC26A4 (7q22.3).
- OMIM®:** 274600.
- Definition:** Pendred syndrome/horsyndrome; enlarged vestibular aqueduct (PDS/NDEVA) comprises a phenotypic spectrum of sensorineural hearing loss (SNHL) that is usually congenital and often severe to profound (although mild-to-moderate progressive hearing impairment also occurs), vestibular dysfunction, and temporal bone abnormalities (distal; enlarged vestibular aqueduct with or without cochlear hypoplasia). PDS also includes development of euthyroid goiter in late childhood to early adulthood whereas NDEVA does not. (From GeneReviews).
- Additional descriptions:** From OMIM: Pendred syndrome, the most common syndromic form of deafness, is an autosomal recessive disorder associated with developmental abnormalities of the cochlea, sensorineural hearing loss, and diffuse thyroid enlargement (goiter) (Livakoff et al., 1997). For a general phenotypic description and a discussion of genetic heterogeneity of thyroid dysgenesis, see TDH1 (274800). <http://www.omim.org/entry/274600>. From ORF: Pendred syndrome is a disorder typically associated with hearing loss and a thyroid condition called a goiter. A goiter is an enlargement of the thyroid gland, which is a butterfly-shaped organ at the base of the neck that produces hormones. If a goiter develops in a person with Pendred syndrome, it usually forms between late childhood and early adulthood. In most cases, this enlargement does not cause the thyroid to malfunction. In most people with Pendred syndrome, severe to profound hearing loss caused by changes in the inner ear (sensorineural hearing loss) is evident at birth. Less commonly, hearing loss does not develop until later in infancy or early childhood. Some affected individuals also have problems with balance caused by dysfunction of the vestibular system, which is the part of the inner ear that helps maintain the body's balance and coordinates an inner ear.
- Go to:** Links to related resources.
- Genetic Testing Registry:** A list of testing options including: Deletion/duplication analysis (16), Microsatellite instability testing (MSI) (1), Mutation scanning of select exons (1), Research (1), Sequence analysis of select exons (4), Sequence analysis of the entire coding region (79), Targeted variant analysis (12), and See all (56).
- Clinical resources:** A link to clinical resources.

一括処理に便利  
閲覧にも便利？

<https://www.ncbi.nlm.nih.gov/medgen/>

疾患（表現型）と遺伝子の  
統合的なデータベース

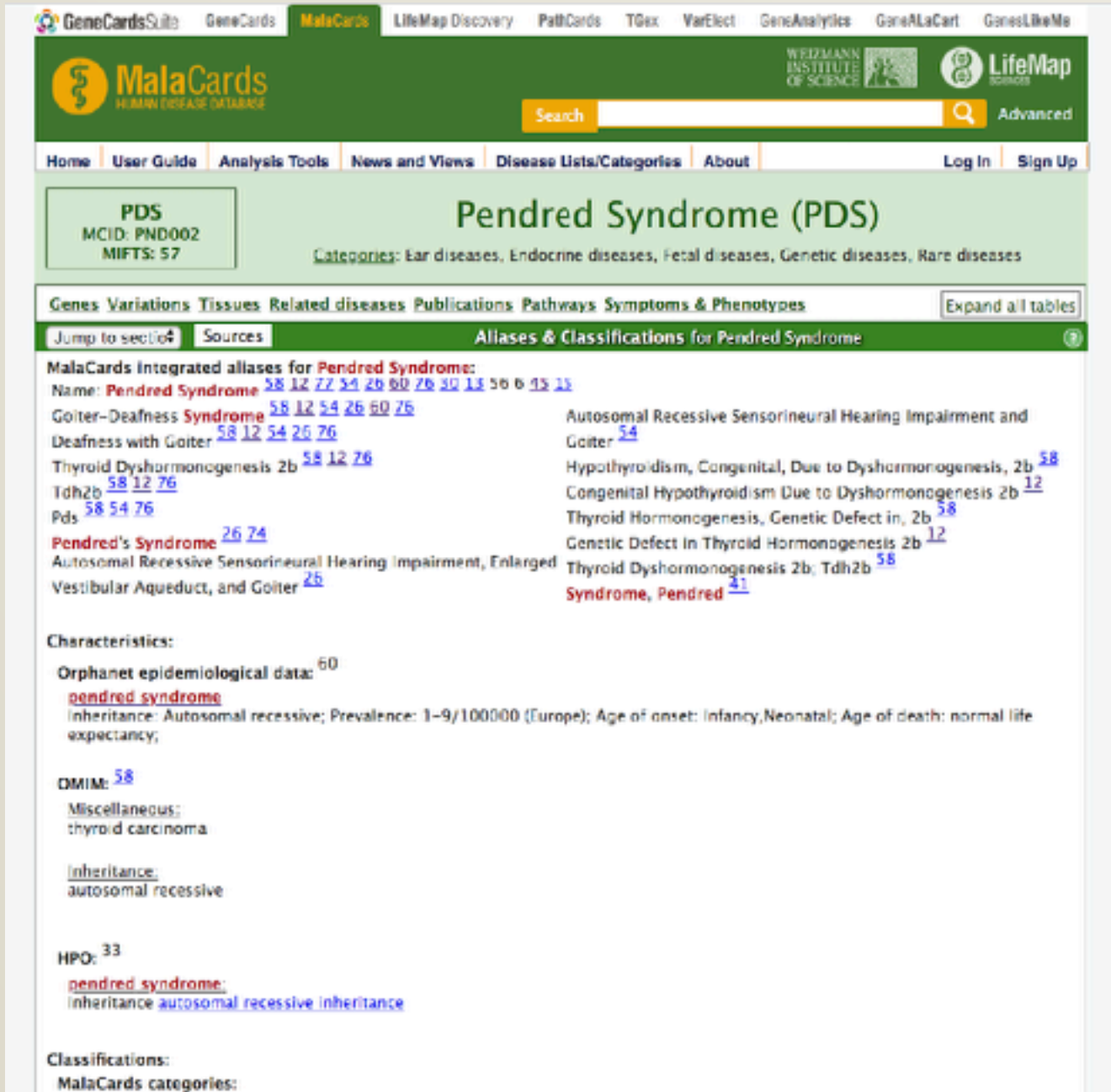
by NCBI

30937 entries

FTPサイトにデータが置かれている

他データベースのエントリとの  
変換テーブルも作成されている  
(OMIM, Gene など)

# MalaCards



The screenshot displays the MalaCards website interface. At the top, there is a navigation bar with links to GeneCards Suite, GeneCards, MalaCards, LifeMap Discovery, PathCards, TGex, VarElect, GeneAnalytics, GeneAlaCart, and GenesLikeMe. The MalaCards logo is prominently displayed on the left, and the Weizmann Institute of Science and LifeMap Sciences logos are on the right. A search bar is located in the center of the top bar. Below the navigation bar, a horizontal menu contains links to Home, User Guide, Analysis Tools, News and Views, Disease Lists/Categories, and About. Log In and Sign Up buttons are on the right. The main content area features a green header for 'Pendred Syndrome (PDS)' with its MCID (PND002) and MFTS (57). Below this, a list of categories includes Ear diseases, Endocrine diseases, Fetal diseases, Genetic diseases, and Rare diseases. A tabbed interface allows users to view Genes, Variations, Tissues, Related diseases, Publications, Pathways, Symptoms & Phenotypes, and an 'Expand all tables' button. A 'Jump to section' dropdown and a 'Sources' link are also present. The 'Aliases & Classifications for Pendred Syndrome' section lists various names and their associated MalaCards IDs, such as 'Pendred Syndrome' (58, 12, 77, 54, 26, 60, 76, 30, 13, 56, 6, 13, 13), 'Goiter-Deafness Syndrome' (58, 12, 54, 26, 60, 76), 'Deafness with Goiter' (58, 12, 54, 26, 76), 'Thyroid Dyshormonogenesis 2b' (58, 12, 76), 'Tdh2b' (58, 12, 76), 'Pds' (58, 54, 76), 'Pendred's Syndrome' (26, 74), 'Autosomal Recessive Sensorineural Hearing Impairment, Enlarged Vestibular Aqueduct, and Goiter' (26), 'Autosomal Recessive Sensorineural Hearing Impairment and Goiter' (54), 'Hypothyroidism, Congenital, Due to Dyshormonogenesis, 2b' (58), 'Congenital Hypothyroidism Due to Dyshormonogenesis 2b' (12), 'Thyroid Hormonogenesis, Genetic Defect in, 2b' (58), 'Genetic Defect in Thyroid Hormonogenesis 2b' (12), 'Thyroid Dyshormonogenesis 2b; Tdh2b' (58), and 'Syndrome, Pendred' (31). The 'Characteristics' section includes Orphanet epidemiological data (60), a link to 'pendred syndrome', inheritance information (Autosomal recessive; Prevalence: 1-9/100000 (Europe); Age of onset: Infancy, Neonatal; Age of death: normal life expectancy), OMIM (58), a link to 'OMIM: 58', a link to 'Miscellaneous: thyroid carcinoma', a link to 'Inheritance: autosomal recessive', HPO (33), a link to 'pendred syndrome', and inheritance information (autosomal recessive inheritance). The 'Classifications' section includes MalaCards categories.

<https://www.malacards.org/>

疾患（表現型）と遺伝子の  
統合的なデータベース

by Weizmann Institute of Science  
(Israel)

19941 entries

GeneCardのシリーズ



# KEGG Disease



## DISEASE: 遺伝性難聴 (常染色体劣性)

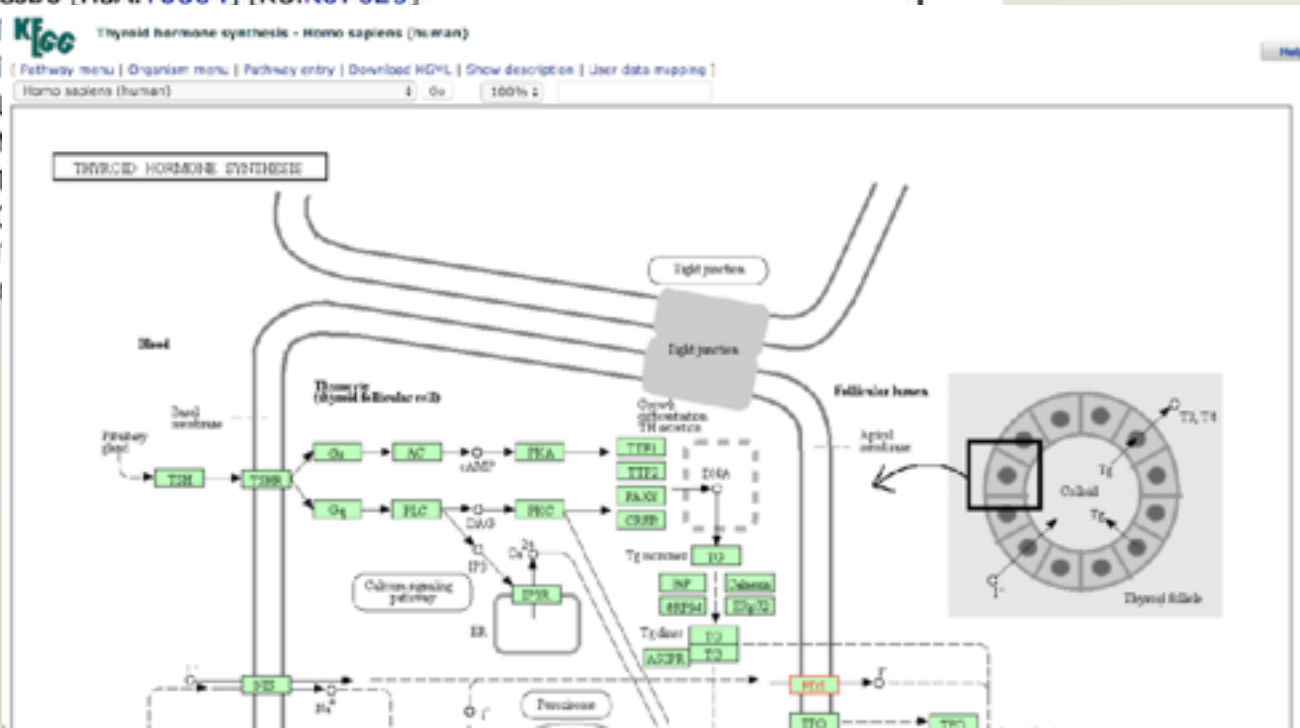
[Help](#)

エントリ	H00605
名称	遺伝性難聴 (常染色体劣性)
概要	遺伝性難聴は、症候型(様々な他の異常を伴う難聴)と非症候型に分けられる。非症候型が遺伝性症例の70%, 症候型が30%を占める。遺伝形式は、常染色体劣性遺伝形式が最もよく見られ(非症候型難聴症例の75%-85%), 次いで常染色体優性遺伝形式(12-13%)とX-連鎖性やミトコンドリア型(2-3%)が続く。常染色体劣性遺伝性難聴は、一般的に最も重症で、専ら蝸牛の欠陥(感音性難聴)に起因する。症候型ではほとんどの症例で伝導性難聴(外耳、中耳の発達異常)または混合型であると対照的である。
カテゴリ	神経系疾患
階層分類	ヒト疾患 [BR:jp08402] 神経系疾患 耳疾患 H00605 遺伝性難聴 (常染色体劣性) ICD-11 による疾患分類 [BR:jp08403] 10 Diseases of the ear or mastoid process Disorders with hearing impairment AB50 Congenital hearing impairment H00605 遺伝性難聴 (常染色体劣性) <a href="#">BRITe hierarchy</a>
病因遺伝子	(DFNB1A) GJB2 [HSA:2706] [KO:K07621] (DFNB1A) GJB3 [HSA:2707] [KO:K07622] (DFNB1B) GJB6 [HSA:10804] [KO:K07625] (DFNB2) M  Thyroid hormone synthesis - Homo sapiens (human) (DFNB3) M <a href="#">Pathway menu</a>   <a href="#">Organism menu</a>   <a href="#">Pathway entry</a>   <a href="#">Download KEGG</a>   <a href="#">Show description</a>   <a href="#">User data mapping</a> (DFNB4) SI (DFNB6) TI (DFNB7/11) (DFNB8/10) (DFNB9) O (DFNB12) I

[https://www.genome.jp/kegg/disease/disease\\_ja.html](https://www.genome.jp/kegg/disease/disease_ja.html)

pathwayから派生した疾患の  
データベース  
(疾患 → 遺伝子 → pathway)

by 京都大・金久研究室





# ClinVar

NCBI Resource | How To | Sign in to NCBI

ClinVar | Search ClinVar | SLC26A4[gene] | Search | Create alert | Advanced

Home | About | Access | Help | Submit | Statistics | FTP

Clinical significance: Conflicting interpretations (23), Benign (60), Likely benign (60), Uncertain significance (190), Likely pathogenic (131), Pathogenic (132), Risk factor (6)

Molecular consequence: Frameshift (32), Missense (200), Nonsense (23), Splice site (18), rRNA (2), Near gene (5), UTR (23)

Variation type: Deletion (64), Duplication (21), Indel (2), Insertion (21), Single nucleotide (360)

Variant length: Less than 51 bp (413), Between 51 and 1000 bp (7), Between 1 and 50 kb (5), Between 50 and 500 kb (3), Between 500 kb and 1 Mb (2), Between 1 and 5 Mb (2), Greater than 5 Mb (5)

Showing results for variants in the **SLC26A4** gene. Search instead for all ClinVar records that mention **SLC26A4**

**Search results**  
Items: 1 to 100 of 461

	Variation Location	Gene(s)	Condition(s)	Clinical significance (if relevant)	Review status
1.	<a href="#">SLC26A4, c.58, C&gt;G, 1002-4</a>	<a href="#">SLC26A4</a>	Pendred syndrome	Pathogenic (Oct 1, 2001)	no assertion criteria provided
2.	<a href="#">SLC26A4, 1-BP INS, 2182G</a>	<a href="#">SLC26A4</a>	Pendred syndrome	Pathogenic (Jun 1, 2006)	no assertion criteria provided
3.	<a href="#">SLC26A4, 1-BP DEL, 1162T</a>	<a href="#">SLC26A4</a>	Pendred syndrome	Pathogenic (Jun 1, 2006)	no assertion criteria provided
4.	<a href="#">SLC26A4, c.254DS, A&gt;G, +2</a>	<a href="#">SLC26A4</a>	Pendred syndrome	Pathogenic (Jun 1, 1999)	no assertion criteria provided
5.	<a href="#">SLC26A4, 5-SP INS, NT2111</a>	<a href="#">SLC26A4</a>	Enlarged vestibular aqueduct	Pathogenic (Jun 1, 1999)	no assertion criteria provided
6.	<a href="#">SLC26A4, 1-BP DEL, 811T</a>	<a href="#">SLC26A4</a>	Enlarged vestibular aqueduct	Pathogenic (Jun 1, 1999)	no assertion criteria provided
7.	<a href="#">SLC26A4, 1-BP DEL, 1621T</a>	<a href="#">SLC26A4</a>	Pendred syndrome	Pathogenic (Dec 1, 1997)	no assertion criteria provided

<https://www.ncbi.nlm.nih.gov/clinvar/>

ヒト variation と疾患（表現型）のデータベース

by NCBI

509259 entries

NCBI Resource | How To | Sign in to NCBI

ClinVar | Search ClinVar | Search ClinVar for gene symbols, HGVS expressions, conditions, and more | Search | Advanced | Help

Home | About | Access | Help | Submit | Statistics | FTP

**NEW** Click here to see the new Variation Report design!

**SLC26A4, 1-BP INS, 2182G**

Variation ID: 4820  
Review status: 0/4 no assertion criteria provided

**Interpretation** Go to: [icon]

Clinical significance: **Pathogenic**  
Last evaluated: Jul 1, 2000  
Number of submission(s): 1  
Condition(s): Pendred syndrome [MedGen - OMIM - OMIM]  
See supporting ClinVar records [2]

**Allele(s)** Go to: [icon]

**SLC26A4, 1-BP INS, 2182G**

Allele ID: 12871  
Variant type: Insertion  
Cytogenetic location: 7q31  
Other names: - 1-BP INS, 7,820

**1 Affected gene**  
cystic carrier family 26 member 4 [SLC26A4] [Gene - OMIM]  
Search ClinVar for variants within SLC26A4  
Search ClinVar for variants including SLC26A4

**Variant frequency in dbGaP**  
No dbGaP data has been submitted for this variant.

**Related information**  
Functional Class  
Gene  
MedGen  
OMIM  
PubMed  
Related genes (specific)

# Orphanet

orphanet pendred Search

Help Print Contact us EN

## Pendred syndrome [Suggest an update](#)

**Disease definition**

Pendred syndrome (PDS) is a clinically variable genetic disorder characterized by bilateral sensorineural hearing loss and euthyroid goiter.

**ORPHA:705**

Synonym(s):	Age of onset: Infancy, Neonatal	MeSH: C536648
Goiter-deafness syndrome	ICD-10: E07.1	GAIRD: 4271
Prevalence: 1-9/100 000	OMIM: 274600	MedDRA: -
Inheritance: Autosomal recessive	UMLS: C0271829	

### Summary

#### Epidemiology

PDS is one of the most frequent forms of syndromic genetic deafness. Although prevalence is unknown, PDS may account for up to 7.5% of cases of congenital hearing loss.

#### Clinical description

Considerable phenotypic variability is found even within families. The main presenting clinical sign is prelingual sensorineural deafness, although occasionally the hearing loss develops later in childhood. The degree of hearing loss is

<https://www.orpha.net/>

希少疾患とorphaned drugの  
データベース

Orphanet コンソーシアム  
(仏・Insermが中心)

5856 diseases and  
3573 genes

# PubCaseFinder



<https://pubcasefinder.dbcls.jp/>

症状から希少疾患を検索する  
ウェブサービス

by DBCLS

4066 rare diseases,  
4002 disease-gene associations,  
1M+ case reports

# HPO (Human Phenotype Ontology)

human phenotype ontology

Tools Downloads Help

All Search for phenotypes, diseases

Pendred Syndrome OMIM:274600

Export Associations

HPO Associations Gene Associations

Ear [ 3 annotations ]

Term Identifier	Term Name	Definition
HP:001751	Vestibular dysfunction	An abnormality of the functioning of the vestibular apparatus.
HP:000527	Congenital sensor neural hearing impairment	A type of hearing impairment caused by an abnormal functionality of the cochlear nerve with congenital onset.
HP:0008754	Cochlear malformation	The presence of a malformed cochlea.

Endocrine [ 2 annotations ]

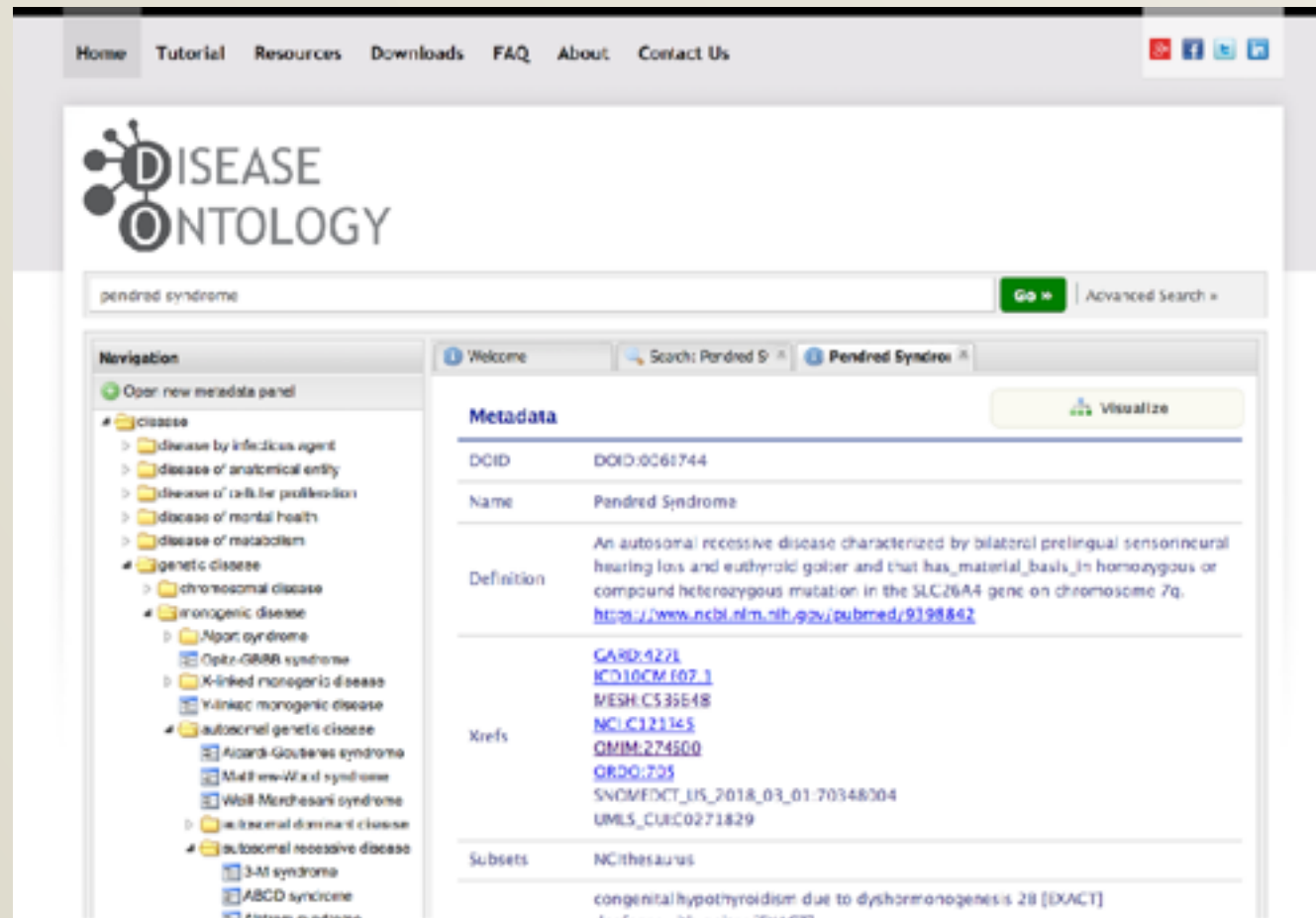
Term Identifier	Term Name	Definition
HP:0000853	Goiter	An enlargement of the thyroid gland.

<https://hpo.jax.org/app/>

ヒト表現型のオントロジー

遺伝子や疾患に対して  
対応づけられた情報も

# DO (Disease Ontology)



<http://disease-ontology.org/>

ヒト疾患のオントロジー



# 疾患データベースの活用例：Gendoo

## Disease

→ [Tree View](#)

	MeSH term	Japanese	Link	01	02
<input type="checkbox"/>	<a href="#">Diabetes Mellitus, Type 1</a>	糖尿病-1型	<a href="#">MeSH</a> <a href="#">LSDB</a>		
<input type="checkbox"/>	<a href="#">Diabetes Mellitus, Type 2</a>	糖尿病-2型	<a href="#">MeSH</a> <a href="#">LSDB</a>		
<input type="checkbox"/>	<a href="#">Insulin Resistance</a>	インスリン抵抗性	<a href="#">MeSH</a> <a href="#">LSDB</a>		
<input type="checkbox"/>	<a href="#">Diabetes Mellitus</a>	糖尿病	<a href="#">MeSH</a> <a href="#">LSDB</a>		
<input type="checkbox"/>	<a href="#">Genetic Predisposition to Disease</a>	遺伝的素因(疾患)	<a href="#">MeSH</a> <a href="#">LSDB</a>		
<input type="checkbox"/>	<a href="#">Obesity</a>	肥満	<a href="#">MeSH</a> <a href="#">LSDB</a>		
<input type="checkbox"/>	<a href="#">Disease Susceptibility</a>	疾病感受性	<a href="#">MeSH</a> <a href="#">LSDB</a>		
<input type="checkbox"/>	<a href="#">Diabetes Mellitus, Experimental</a>	糖尿病-実験的	<a href="#">MeSH</a> <a href="#">LSDB</a>		
<input type="checkbox"/>	<a href="#">Hyperglycemia</a>	高血糖症	<a href="#">MeSH</a> <a href="#">LSDB</a>		
<input type="checkbox"/>	<a href="#">Prediabetic State</a>	前糖尿病状態	<a href="#">MeSH</a> <a href="#">LSDB</a>		
<input type="checkbox"/>	<a href="#">Syndrome</a>	症候群	<a href="#">MeSH</a> <a href="#">LSDB</a>		
<input type="checkbox"/>	<a href="#">Body Weight</a>	体重	<a href="#">MeSH</a> <a href="#">LSDB</a>		
<input type="checkbox"/>	<a href="#">Glucose Intolerance</a>	耐糖能障害	<a href="#">MeSH</a> <a href="#">LSDB</a>		
<input type="checkbox"/>	<a href="#">Autoimmune Diseases</a>	自己免疫疾患	<a href="#">MeSH</a> <a href="#">LSDB</a>		
<input type="checkbox"/>	<a href="#">Pancreatic Diseases</a>	膵臓疾患	<a href="#">MeSH</a> <a href="#">LSDB</a>		
<input type="checkbox"/>	<a href="#">Diabetic Ketoacidosis</a>	糖尿病性ケトアシドーシス	<a href="#">MeSH</a> <a href="#">LSDB</a>		

疾患をMeSHキーワードを用いてプロファイリング

# データ作成の流れ

Step 1: Extract PMIDs cited in Entrez Gene/OMIM reference sections



Reference section

OMIM ID

PMID

Step 2: Collect Entrez Gene/OMIM IDs described in MEDLINE articles



OMIM ID

PMID

Gene/OMIM-  
PMID Pairs

Gene/OMIM-  
MeSH Pairs

Step 3: Collect PMIDs assigned by MeSH terms corresponding to genes and diseases



Corresponding  
MeSH terms

OMIM ID

PMID

PubMed  
Search

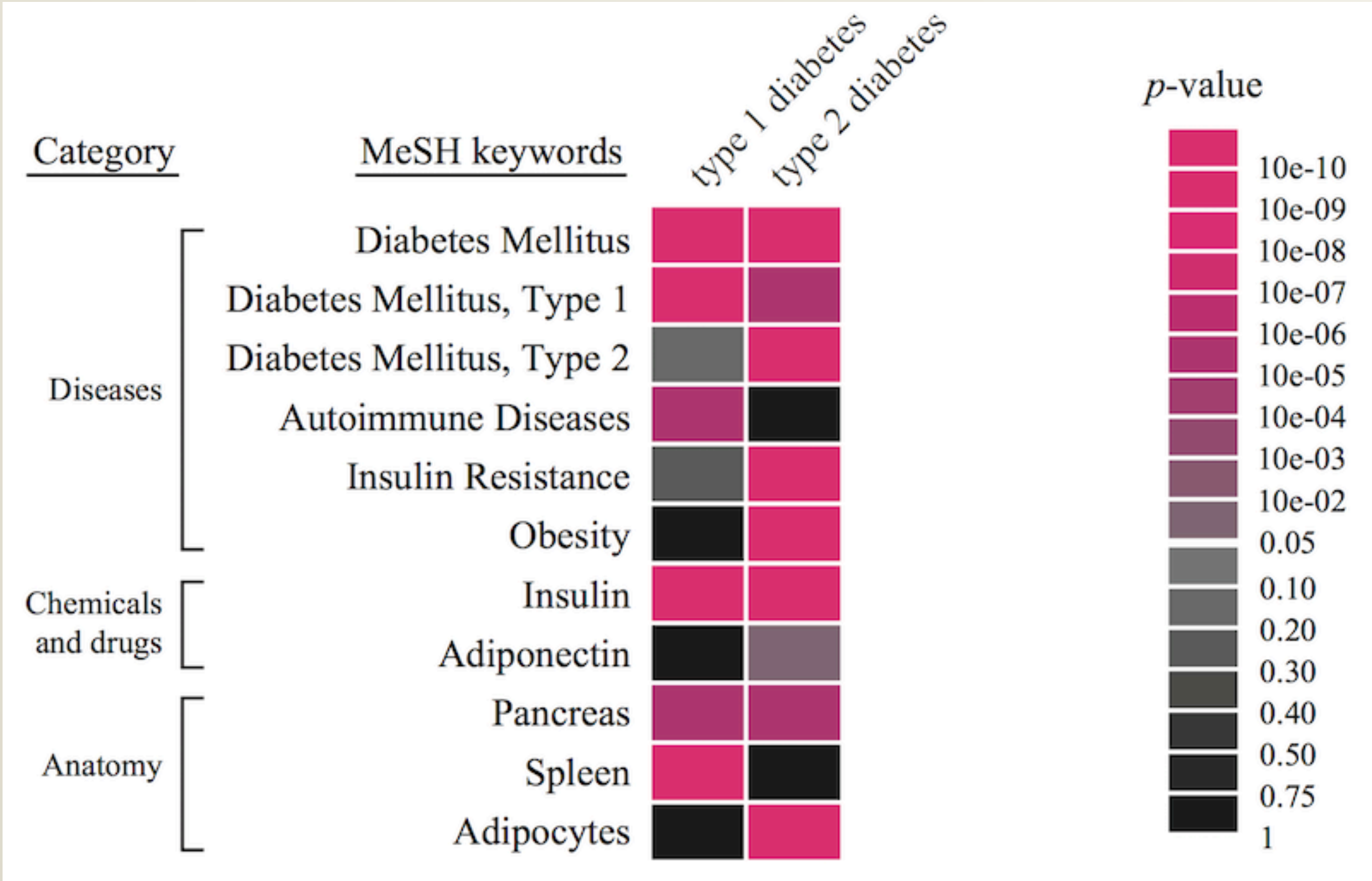
Scoring

p-value



# 疾患データの外部データとの連携例

例：1型/2型糖尿病のプロファイルの違い





# 別の例：疾患関連NGSデータの検索

Disease Type:

Total: 303 << first < prev 1 2 3 4 5 6 7 8 9 10 next > last >> 10

Disease	疾患名	# of submission
Breast Neoplasms	乳房腫瘍	43
Prostatic Neoplasms	前立腺腫瘍	22
Disease Models, Animal	疾患モデル(動物)	21
Genetic Predisposition to Disease	遺伝的素因(疾患)	
Disease Progression	病勢悪化	
Translocation, Genetic	転座	
Cell Transformation, Neoplastic	腫瘍細胞形質転換	
Lung Neoplasms	肺腫瘍	
Staphylococcal Infections	ブドウ球菌感染症	
Malaria	マラリア	

Total: 303 << first < prev 1 2 3 4 5 6 7

Total: 6 << first < prev 1 next > last >> 10

SRA ID	SRA Title	Disease	疾患名	PMID
SRA026055	DNA sequencing of a cytogenetically normal acute myeloid leukaemia genome	Leukemia, Myeloid, Acute	白血病-急性骨髄性	18987736
SRA026055	DNA sequencing of a cytogenetically normal acute myeloid leukaemia genome	Leukemia, Myeloid, Acute	白血病-急性骨髄性	19657110
SRA026055	Recurring Mutations Found by Sequencing an Acute Myeloid Leukemia Genome	Leukemia, Myeloid, Acute	白血病-急性骨髄性	18987736
SRA026055	Recurring Mutations Found by Sequencing an Acute Myeloid Leukemia Genome	Leukemia, Myeloid, Acute	白血病-急性骨髄性	19657110
SRA009897	In-depth characterization of the microRNA transcriptome in a leukemia progression model	Leukemia, Myeloid, Acute	白血病-急性骨髄性	18849523
SRA029797	Exome Sequencing Identifies Somatic Mutations in Acute Monocytic Leukemia	Leukemia, Myeloid, Acute	白血病-急性骨髄性	21399634

Total: 6 << first < prev 1 next > last >> 10

# 参考リソース



# 参考図書・その1 ～ ウェブツール使い方集



2点すべてのイメージを見る

## 生命科学データベース・ウェブツール 図解と動画で使い方がわかる! 研究がはかどる定番18選 単行本 (ソフトカバー) - 2018/12/4

坊農秀雅 (編集), 小野浩雅 (編集)

カスタマーレビューを書きませんか?

＞ その他 ( ) の形式およびエディションを表示する

単行本 (ソフトカバー)

¥ 3,024

¥ 5,362 より 4 中古品の出品  
¥ 3,024 より 3 新品

5/25 土曜日 にお届けするには、今から**21 時間 4 分**以内にお急ぎ便を選択して注文を確定してください (Amazonプライム会員は無料 [詳細を見る](#))

まとめ買いで **最大10%** ポイント還元 【2冊で最大4%、3冊以上で最大8%、10冊以上で最大10%】

2冊を購入する際クーポンコード「2BOOKS」を、3冊以上は「MATOME」を入力すると最大8~10%ポイント還元! [今すぐチェック](#)

シェアする    

¥ 3,024

ポイント: 30pt (1%)


[詳細はこちら](#)

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残り**13点** (入荷予定あり) [在庫状況について](#)

この商品は、[Amazon.co.jp](#) が販売、発送します。

数量:

 カートに入れる

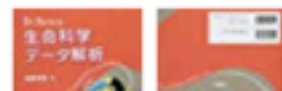
 今すぐ買う

時間を節約

1-Click注文を有効にすると、すぐに注文できます。

[詳細はこちら](#)

# 参考図書・その2 ～ 初心者向けバイオインフォ本



2点すべてのイメージを見る

## Dr. Bonoの生命科学データ解析 単行本 - 2017/9/29

坊農秀雅 (著)

★★★★★ 3件のカスタマーレビュー

その他 ( ) の形式およびエディションを表示する

単行本  
¥ 3,240

¥ 4,205 より 8 中古品の出品  
¥ 3,240 より 4 新品

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まとめ買いで **最大10%** ポイント還元  
【2冊で最大4%、3冊以上で最大8%、10冊以上で最大10%】ポイント還元  
2冊を購入する際クーポンコード「2BOOKS」を、3冊以上は「MATOME」を入力すると最大8~10%ポイント還元！ [今すぐチェック](#)

シェアする    

¥ 3,240

ポイント: 96pt (3%)

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数量:

 カートに入れる

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時間を節約

1-Click注文を有効にすると、すぐに注文できます。

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