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Answer to GUDMAP Exercise Question 5:

Which genes are associated with Human renal tubular dysgenesis and what are their chromosomal locations?

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GUDMAP Web Portal

www.gudmap.org

Select Disease →

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Database Statistics

Assay Type	Entries	Genes
All In Situ Hybridisation (ISH):	10758	3692
Wholemout ISH (WISH):	7288	2896
Section ISH (SISH):	3406	1436
OPT ISH:	64	32
Immunohistochemistry (IHC):	326	20
Transgenic Reporters:	41	28
Microarray:	461	-
More ...		

Search Data

Marker Mouse Strains

NEW
Reporter Strain Nominations

The **GenitoUrinary Development Molecular Anatomy Project** (GUDMAP) is a consortium of laboratories working to provide the scientific and medical community with tools to facilitate research. The key components are:

- a molecular atlas of gene expression for the developing organs of the GenitoUrinary (GU) tract
- a high resolution molecular anatomy that highlights development of the GU system
- mouse strains to facilitate developmental and functional studies within the GU system
- tutorials describing GU organogenesis
- rapid access to primary data via the GUDMAP database

The GUDMAP tools, web site and database are a public resource funded by the National Institutes of Health, USA.

Web Demos Download Data Development Tutorials

Developmental Cell
Gendhi et al. *Development and Organogenesis*
Dev Cell, 2013 Sep 16;26(5):469-82.

Image use policy


GUDMAP
GenitoUrinary Molecular Anatomy Project

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GUDMAP Disease Resource

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[Query Disease-Gene Associations](#)  **Select "Disease-Gene Associations"**
[Query Phenotype-Gene Associations](#)

The [GATACA GUDMAP Gene Explorer](#) at Cincinnati Children's Hospital Medical Center
A tool for the identification and analysis of genes responsible for functions and diseases of the genitourinary system.


[ToppGene Suite](#) at Cincinnati Children's Hospital Medical Center
A one-stop portal for gene list enrichment analysis and candidate gene prioritization based on functional annotations and protein interaction networks.

Querying GUDMAP Disease-Gene Associations

How are disease-gene associations determined?

To search for **genes associated with a genitourinary disease**:

Select the OMIM genitourinary disease of interest from the pull-down menu. Select your disease of interest and click 'Search'. Alternatively, enter the disease name or term you wish to search for in the text box. You can enter any term you like and the query will search for all OMIM disease names that contain your search term (e.g. entering 'wilms' will return all 'Wilms Tumor' entries). Bear in mind, if using the pull-down menu be sure to keep the text box empty as this will take precedence. The query will bring back a table of genes that are associated with the chosen disease.

Select Disease:  **Select disease from drop-down menu**

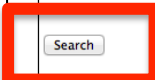
Enter Disease:

To search for **genes associated with a disease**:

The pull-down menu is used to select a gene of interest. Enter the gene name in the text box. You can enter any term you like and the query will search for all OMIM disease names that contain your search term (e.g. entering 'wilms' will return all 'Wilms Tumor' entries). Bear in mind, if using the pull-down menu be sure to keep the text box empty as this will take precedence. The query will bring back diseases that are associated with the chosen gene.

Select Gene:

Enter Gene:

 **Select "Search"**


GUDMAP Disease Resource - Query Results

Results of query for genes associated with OMIM DISEASE: **RENAL TUBULAR DYSGENESIS**


- Click on the gene symbol to go to the gene page within the GUDMAP gene expression database.
- **Please note:** Some genes may not have a gene page within GUDMAP.
- Click on the OMIM ID to go the disease page at OMIM - the nature of the association can be checked here.
- The *In Situ Data* column indicates whether there is available in situ data in the GUDMAP gene expression for that gene.
'1' = yes, '0' = no.
- The *NCBI* column if there is evidence supporting the association from NCBI.

Records 1 to 4 of 4

OMIM ID	Disease Name	Human Gene Symbol	Mouse Gene Symbol	Mouse Gene MGI ID	In Situ Data
267430	RENAL TUBULAR DYSGENESIS	ACE	Ace	MGI:87874	yes
267430	RENAL TUBULAR DYSGENESIS	AGT	Agt	MGI:87963	yes
267430	RENAL TUBULAR DYSGENESIS	AGTR1	Agtr1a	MGI:87964	yes
267430	RENAL TUBULAR DYSGENESIS	REN	Ren1	MGI:97898	yes



Click here for OMIM entry
containing chromosomal
location



Human genes associated
with renal tubular
dysgenesis

OMIM entry for renal tubular dysgenesis

#267430 ICD+

RENAL TUBULAR DYSGENESIS; RTD

Alternative titles; symbols
PRIMITIVE RENAL TUBULE SYNDROME

Other entities represented in this entry:
RENAL TUBULAR DYSGENESIS WITH CHOANAL ATRESIA AND ATHELIA, INCLUDED

Phenotype-Gene Relationships

Location	Phenotype	Phenotype MIM number	Phenotype mapping key	Gene/Locus	Gene/Locus MIM number
1q32.1	Renal tubular dysgenesis	267430	3	REN	179820
1q42.2	Renal tubular dysgenesis	267430	3	AGT	106150
3q24	Renal tubular dysgenesis	267430	3	AGTR1	106165
17q23.3	Renal tubular dysgenesis	267430	3	ACE	106180

[Clinical Synopsis](#)

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External Links for Entry:

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Centers for Mendelian Genomics

**Chromosomal location of
Human genes associated
with renal tubular
dysgenesis**

- Question 5: Which genes are associated with Human renal tubular dysgenesis and what are their chromosomal locations?
- Answer: ACE (17q23.3), AGT (1q42.2), AGTR1 (3q24) and REN (1q32.1).