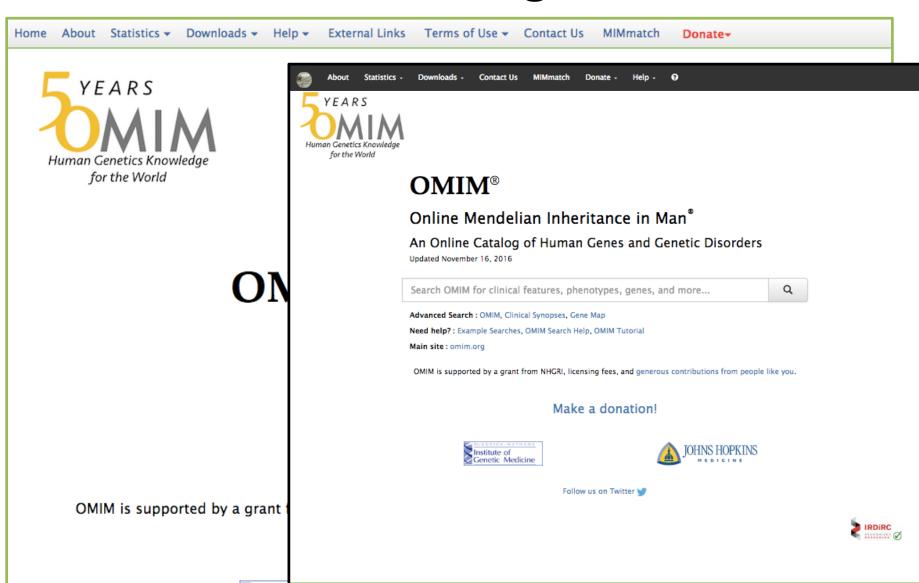
## OMIM®

Ada Hamosh, MD, MPH Scientific Director

## **OMIM.org**



NOTE: OMIM is intended for use primarily by physicians and other professionals concerned with genetic disorders, by genetics researchers, and by advanced students in science and medicine. While the OMIM database is open to the public, users seeking information about a personal medical or genetic condition are urged to consult with a qualified physician for diagnosis and for answers to personal questions.

### **OMIM**

- Knowledgebase of human genes and genetic phenotypes
- >15,400 genes, and >8,000 phenotypes
- >26,000 selected variants showing gene-phenotype relationships
- Structured free-text descriptions for human consumption
  - Loaded into an SQL database with up to 58 Boolean fields and meta-tagged text
- Clinical Synopses, Phenotypic Series, and Gene Map
- Robust REST API for computational queries
- Copious topical links to 48 external data resources, all indexed and available through the API

### OMIM's Focus

- Name and describe new Mendelian phenotypes as well as those with significant genetic contribution
- Describe Gene-Phenotype Relationships
- Describe the genes that cause disease
- Describe other genes of known function

- OMIM is based on the peer-reviewed biomedical literature
  - Expert MD and PhD science writers abstract important new information to add. All information is reviewed and entered by an experienced biocurator. The selection process is key.

#### **OMIM Staff**

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Deputy Scientific Director for Phenotypes: Nara Sobreira, MD, PhD

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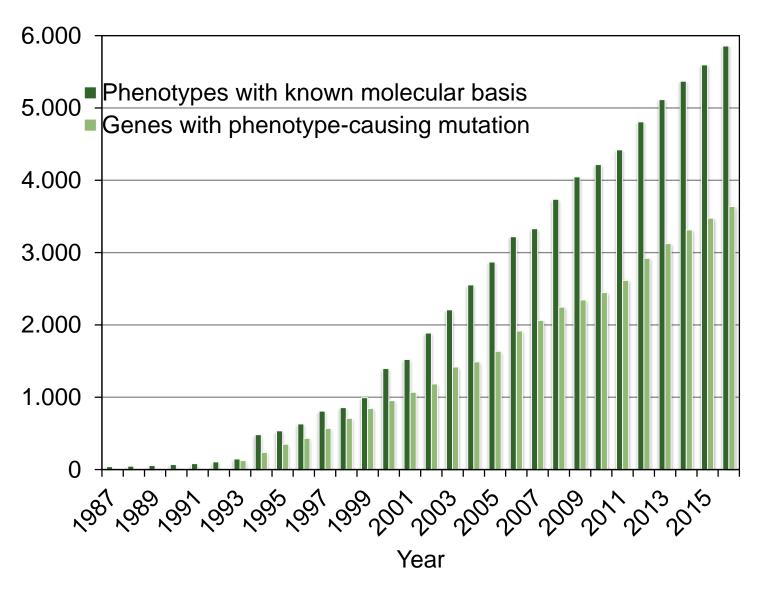
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#### Growth of Gene-Phenotype Relationship

12 October 2016



Source: Online Mendelian Inheritance in Man

## **OMIM USAGE**

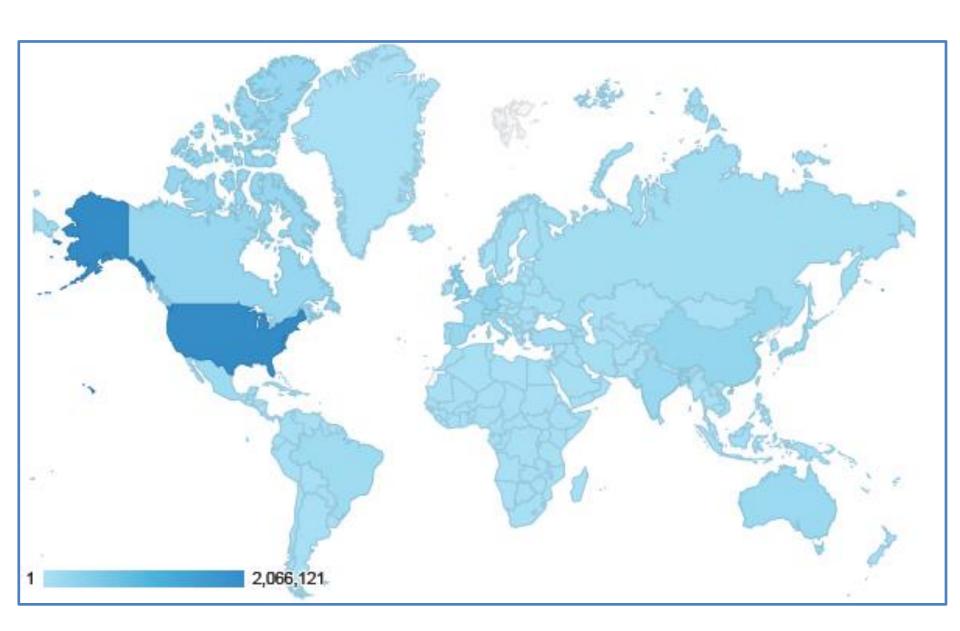
# Monitoring of Use and Usefulness

- Periodic User Surveys
  - Most recent pop-up from the website for 2 weeks in April 2016
  - 5409 responses: 56% use it ≥weekly, 12% first time
  - "What work takes up most of your time?"
    24% clinical medicine, 23% basic science,
    12% clinical lab, 8% bioinformatics
  - 39% of respondents were students

# OMIM Usage past 12 months

- >2.4 million unique users, with >6 million sessions, >26 million page views
- >4 page views on average in 5.5 minutes per session
- 60-90,000 users per week
- >785,000 unique users accessed >100 times in the year
- 54% of use is from the U.S.

# OMIM: Worldwide Use Nov 2015-Oct 2016



# OMIM Use Statistics from Google Analytics

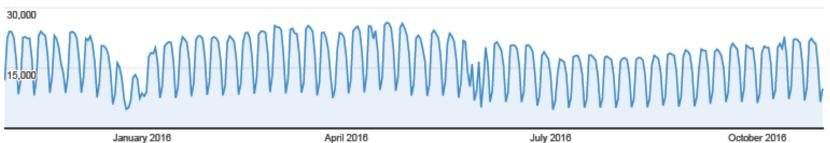


Nov 1, 2015 - Oct 30, 2016



Overview

#### Sessions



Sessions

6,008,426

Users

2,428,446

Pageviews

26,759,564

Pages / Session

4.45

Avg. Session Duration

00:05:48

Bounce Rate

48.86%



% New Sessions

38.56%

### Downloads and API Users

- 47,593 downloads since January 2016
- 2,513 registered API users
- ~80,000 API requests per day
- Peak of >200,000

# **Funding**

- 1966-1978 Single author (Dr. McKusick), funded through book sales
- 1979-1987 Knowledge base program of NLM
- 1987-1994 HHMI/GDB
- 1995-1999 Grant from NHGRI/NCBI
- 1999-2009 Contract from NCBI
- 2010-2016 Grants (1-2.5 years at a time)
   from NHGRI

# **Current Funding Model**

- Grant submitted to NHGRI and recently reviewed
- Since January 2016, API requires registered access. Commercial users must obtain and pay a license fee. Academic, Government, Non-Profits free.
  - >60 licenses in place (over 150 new referred to Technology Ventures), current income <1/3 operating costs
  - Requesting donations from users
  - >800 donations

# International Funding Mechanism

- Certain resources are foundational and essential for research
- Need a mechanism for all nations whose citizens benefit from these resources to support their long-term sustainability
- Possibly a UN- or WHO-based funding mechanism?

# Thank you!

ahamosh@jhmi.edu or

Contact us through OMIM.org

#### Frequency & Recency



#### Distribution

Count of Sessions

Sessions

6,008,426

% of Total: 100.00% (6,008,426)

Pageviews

26,759,564

% of Total: 100.00% (26,759,564)

Count of Sessions	Sessions	Pageviews
1	2,320,613	5,921,652
2	579,300	2,020,874
3	301,670	1,170,832
4	199,169	831,182
5	146,963	645,794
6	115,659	528,240
7	95,016	442,869
8	80,428	386,895
9-14	323,508	1,637,687
15-25	321,425	1,822,188
26-50	379,554	2,427,927
51-100	359,727	2,503,917
101-200	321,861	2,463,843
201+	463,533	3,955,664

#### Growth of the Number of Entries in Mendelian Inheritance in Man

