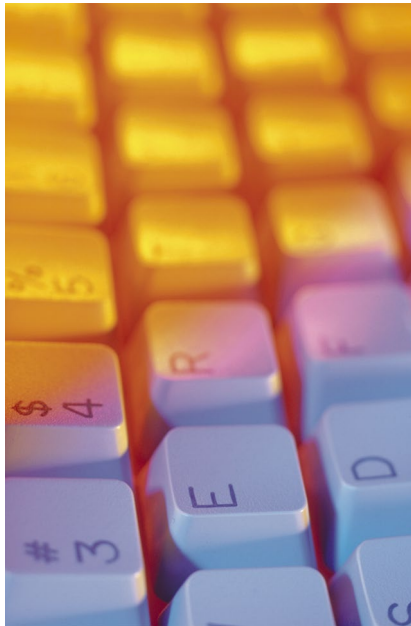


DISEASE GENETICS

Understanding genetic disease with electronic medical records

Science **359**, 1233–1239 (2018)

Cell **173**, 1–13 (2018)



Credit: Aaron Haupt/Science Source

Electronic medical records can be used to better understand heritability patterns of genetic disease, to identify next of kin using emergency contact information, and to uncover previously unrecognized Mendelian disease patterns by leveraging stored phenotypes.

A team from Vanderbilt University mined the electronic medical records of individuals admitted to the university's medical center

to identify phenotypic features that occur in individuals with Mendelian disease. Applying this knowledge to genotyped individuals affected with overlapping symptoms allowed them to identify rare pathogenic mutations causing these diseases.

In another study at three New York-based hospitals, researchers linked family members using emergency contact information, allowing them to accurately trace genetic disease inheritance patterns.

Both studies illustrate the wealth of information contained within electronic health records.

HS

<https://doi.org/10.1038/s41591-018-0121-8>

NEURODEGENERATION

A pig model of Huntington's disease

Cell **173**, 989–1002 (2018)

Knock-in of the huntingtin gene in pigs recreates key features of Huntington's disease.

Huntington's disease is a severe neurodegenerative monogenetic disease that results in age-dependent selective neuronal loss in affected individuals—a feature that cannot be mimicked in murine genetic models of the disease.

Collaborators from the United States and China used gene editing and somatic nuclear cell transfer to generate pigs expressing the huntingtin gene. Crucially, the added gene was inherited through the germline and resulted in age-dependent selective neuron loss mimicking human disease.

The work highlights the relevance of larger animal models to understanding this disease and developing therapeutics.

HS

<https://doi.org/10.1038/s41591-018-0119-2>

INFECTIOUS DISEASE

Detecting dengue

Nature **557**, 719–723 (2018)

A new framework allows the identification of individuals with subclinical dengue infections.

Many dengue fever infections go unobserved, which hampers the wider understanding of disease risk and the identification of correlates of infection. Researchers analyzed blood samples from a school-aged cohort in Thailand to develop a model of antibody dynamics upon infection with dengue. This allowed them to extrapolate diagnoses in infected individuals without outward symptoms. They found previous infection and maintenance of a certain amount of antibodies in blood increased the probability that a second infection would result in hemorrhagic fever.

The model also helps understanding of vaccine response and can be applied to other infectious diseases.

HS

<https://doi.org/10.1038/s41591-018-0120-9>

CANCER

Predicting colon cancer recurrence

Lancet **391** 2128–2139, 2018

Immunoscore, a measure of the level of immune infiltration in tumors, can predict recurrence of colon cancer.

Measures to predict colon cancer prognosis and recurrence are desperately needed to correctly inform and screen affected patients.

In a study of 2,681 patients from 14 countries, all of whom had stage I–III colon cancer, pathologists used a standardized immunohistochemical analysis approach to identify the number of tumor-infiltrating T cells in patients' tumors and to calculate the immunoscore for each patient. They found that immunoscore could reliably predict the recurrence of colon cancer in these patients after 5 years post treatment.

Immunoscore could potentially be used to predict relapse and prognosis in other tumors.

HS

<https://doi.org/10.1038/s41591-018-0123-6>

Hannah Stower

CLINICAL TRIALS

Green light for data sharing

N. Engl. J. Med. **378**, 2202–2211 (2018)

Ninety-two percent of participants of clinical trials perceive that the benefits of sharing their participant-level data outweigh the negatives.

There is a global trend toward sharing data from clinical trial participants for further analysis. Despite researchers' perception that trial participants are concerned that their privacy may be compromised if they participate in a clinical trial, participants' opinions have not yet been studied.

In a study led by a health law researcher, 771 clinical trial participants from three academic medical centers in the United States were surveyed. Most participants responded that, provided that appropriate safeguards are in place, they are happy to share their data for a wide variety of purposes.

The survey indicates that the fears of clinical trial investigators and sponsors may be unfounded.

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<https://doi.org/10.1038/s41591-018-0122-7>