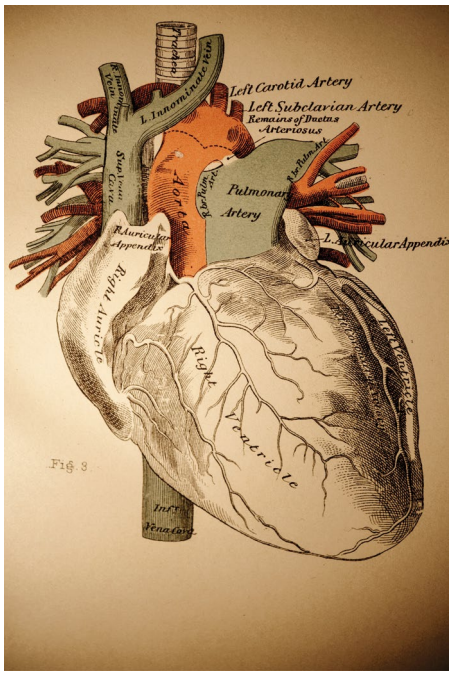
DIAGNOSTIC DEVICES

Identifying atrial fibrillation

JAMA **320**, 146–155 (2018).



Credit: Mark Strozier / Alamy Stock Photo

Atrial fibrillation is the most common sustained arrhythmia, and it substantially increases the risk of stroke in affected individuals. Its diagnosis is important for the correct prescription of anticoagulants to reduce the risk of stroke and is currently made during screening at regular medical checks.

In a clinical trial, participants at high risk for atrial fibrillation wore a self-applied wearable electrocardiogram (ECG) patch that could diagnose this heart disorder. The rate of atrial fibrillation diagnosis and engagement with health care were higher among individuals who wore the device, indicating the effectiveness of these wearable diagnostic devices. *HS*

https://doi.org/10.1038/s41591-018-0287-0

DISEASE GENETICS

Understanding genetic disease with electronic records

Science **359**, 1233–1239 (2018).
Cell **173**, 1–13 (2018).

Electronic medical records contain a wealth of information that can be analyzed to provide insight into disease and disease genetics.

A team from Vanderbilt University mined the electronic medical records of individuals admitted to the university's medical center to develop a score for the aggregate phenotypic features that occur in individuals with Mendelian disease. Applying this knowledge to genotyped individuals affected with overlapping symptoms but undiagnosed disease allowed researchers to identify rare pathogenic mutations causing these diseases.

In another study at three New York–based hospitals, researchers linked family members in their records systems using the emergency contact information provided by patients, allowing the scientists to accurately trace genetic disease inheritance patterns.

Both studies illustrate the importance of maintaining electronic health records and providing researchers with access to them. *HS*

https://doi.org/10.1038/s41591-018-0283-4

SYNTHETIC BIOLOGY

In vivo detection of gut bleeding

Science **360**, 915–918 (2018).

The ability to remotely monitor biomolecules in the gut could allow early diagnosis and monitoring of disease. Thus far, attempts to do this with ingestible biosensors composed of engineered organisms have been hampered by the necessity for complex analysis to interpret information from these biosensors.

A group of researchers from Boston developed an ingestible micro-bio-electronic device (IMBED). The device consists of engineered probiotic bacteria with sensor capabilities combined with low-powered microelectronics that allow remote monitoring of biomolecules in the gut. The scientists were able to show that a heme-sensitive version of this IMBED could detect gastrointestinal bleeding in a pig experiment. They were also able to combine this sensor with others, providing groundwork for gastrointestinal biomarker discovery and transformation of disease management using this device. *HS*

https://doi.org/10.1038/s41591-018-0289-y

RECOMBINANT PROTEIN THERAPY

In utero correction of a genetic disorder

N. Engl. J. Med. **378**, 1604–1610 (2018).

The genetic disorder X-linked hypohidrotic ectodermal dysplasia (XLHED) results in a lack of the protein ectodysplasin A (EDA) in affected individuals. This prevents the development of sweat glands, which can lead to life-threatening hyperthermia.

Researchers from the University of Erlangen injected the receptor-binding domain of EDA into the amniotic cavity of two pregnant women, one with a single fetus and one with identical twins. The genotypes of these fetuses were unknown, but they were known to be affected by XLHED because of results from noninvasive screening and because they have affected siblings. Once born, the three infants were able to sweat normally.

Although long-term follow-up has yet to be carried out, this study shows the effectiveness of prenatal protein therapy for this genetic disease at critical time periods in development. *HS*

https://doi.org/10.1038/s41591-018-0288-z

IMMUNOTHERAPY

The microbiome influence

Science **359**, 91–97 (2018).
Science **359**, 97–103 (2018).
Science **359**, 104–108 (2018).

In the form of immunotherapy known as immune checkpoint inhibition, therapies are administered that release the breaks (i.e., checkpoints) on the immune system to activate it against cancer. This approach has had a major impact on cancer treatment, but not all individuals treated with this therapy respond.

Researchers from the United States and France found that the composition of the gut microbiome can influence an individual's response to immune checkpoint inhibition. The researchers in France found that antibiotic treatment inhibits the effectiveness of this kind of immunotherapy treatment, and the US scientists found that patients with melanoma who did not respond to immune checkpoint inhibition drugs had an imbalance of gut flora, hampering their immune response against cancer.

In the future, the composition of the gut microbiome could be manipulated to enhance the effectiveness of immune checkpoint inhibition or used as a biomarker to gauge potential response. *HS*

https://doi.org/10.1038/s41591-018-0285-2

Hannah Stower