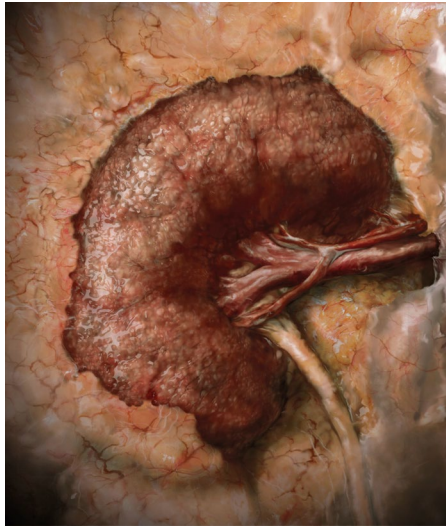


KIDNEY DISEASE

Exome sequencing for chronic kidney disease diagnosis

N. Engl. J. Med. **380**, 142-151 (2019)



Credit: TheVisualMD/Science Source

Exome sequencing is shown to be of clinical utility for diagnosing chronic kidney disease.

Chronic Kidney disease can be difficult to detect via conventional clinical measures, and the inability of doctors to diagnose the disease at early stages has impeded early detection and targeted interventions. Furthermore, the clinical utility of exome sequencing as a first-line diagnostic tool in many diseases has not been systematically assessed.

Researchers from the United States, United Kingdom and Sweden sequenced 3,315 individuals, of which 1,179 were of non-European ancestry, who had conditions that represented all major types of nephropathy.

The researchers found diagnostic variants in 9.3% of the patients, encompassing 66 different monogenic disorders. Notably, those that had the same genetic disorders often presented different clinical symptoms. *HS*

<https://doi.org/10.1038/s41591-019-0361-2>

INFECTIOUS DISEASE

Sequencing for rapid response to Lassa fever

Science **363**, 74-77 (2019)

Samples from the 2018 Nigerian Lassa fever outbreak were sequenced, and results indicated that it was a result of multiple zoonotic transmission events.

Lassa fever is an acute viral hemorrhagic fever that is contracted through exposure to infected rodents or, less frequently, through contact with the bodily fluids of infected humans. In early 2018, the outbreak was seen as unusually large, and there were fears of human-to-human transmission.

An international team applied Nanopore metagenomic sequencing to 120 Lassa-positive samples at the epicenter of the outbreak. Their analyses indicated that the outbreak was the result of multiple zoonotic transmission events. These results were communicated immediately, allowing for alteration of public health policy. *HS*

<https://doi.org/10.1038/s41591-019-0363-0>

NEURODEGENERATION

Transmission of Aβ seeds

Nature **564**, 415-419 (2018)

The Alzheimer's disease-associated protein, amyloid-β protein (Aβ), is transmissible by iatrogenic transmission and can seed deposits in the brain of recipients.

Individuals treated with human cadaveric growth hormone (c-hGH) contaminated with prions during childhood who subsequently developed Creutzfeldt-Jakob disease were previously found to have Aβ deposits in their brains, and had they lived long enough that they may have developed disease.

Researchers from the United Kingdom and Japan were able to track down the c-hGH samples used in these individuals and found that they contained Aβ and tau proteins and that these were able to seed Aβ plaques in mice.

The authors' findings have implications for medical and surgical procedures to try to minimize risk of Aβ transmission. *HS*

<https://doi.org/10.1038/s41591-019-0364-z>

PERSONALIZED MEDICINE

Personalizing metabolic disease therapies

Cell Stem Cell **24**, 1-10 (2019)

Modeling the responses to the insulin-resistance-reversing thiazolidinedione (TZD) drugs in human adipose stem cell (hASC)-derived adipocytes identifies genetic variants that explain patient responses.

TZD drugs are able to reverse insulin resistance, preventing development of type 2 diabetes. However, side effects and variable responses to these drugs inhibit their widespread use.

Scientists from the United States and China derived adipocytes from the adipose stem cells of five obese patients. TZD drugs function by modifying the action of the transcription factor peroxisome proliferator activated receptor γ (PPARγ), and they were able to identify genetic variants that modified the effect of TZD drugs on this protein and hence its effectiveness.

Screening for these variants in the future could tailor therapies. *HS*

<https://doi.org/10.1038/s41591-019-0365-y>

Hannah Stower

ADDICTION

Detecting opioid overdoses with smartphones

Sci. Transl. Med. **11**, eaau8914 (2019)

A system that converts a smartphone into a contactless sensor can provide an early warning to friends or emergency medical services of an individual experiencing an accidental opioid overdose.

Death from accidental opioid overdose can be rapidly prevented if detected early and naxolone is administered. High doses of opioids can cause cessation of breathing (apnea) and respiratory failure.

Researchers from the University of Washington developed an algorithm that could convert the smartphone's native speaker and microphone into a short-range active sonar that is able to detect the early overdose signs. They show it is effective in both simulated overdose situations and a supervised injection facility, indicating that it has the potential to save lives. *HS*

<https://doi.org/10.1038/s41591-019-0362-1>