Breakthroughs Put Diseases On The Back Foot

By James Gallagher
Health and science correspondent, BBC News 24 December 2017
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Peter has Huntington's disease and his siblings Sandy and Frank also have the gene.

It has been a remarkable year of promise in medical science. Incurable diseases from sickle cell to haemophilia (spelled hemophilia in U.S.) now look as though they can be treated. Here are the highlights.

Huntington's

The defect that causes the devastating degenerative disease Huntington's has been corrected in patients for the first time.

It has been called the biggest breakthrough in neurodegenerative diseases for 50 years. The disease is caused by an aberration in a section of DNA called the huntingtin gene. The error corrupts a healthy protein and turns it into a killer of brain cells. The therapy silences the gene by blocking the messages it sends to the cell's protein-making factories.
The study has generated a huge amount of excitement. It could be the first treatment to slow or prevent any degenerative brain disease.

New Skin

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Hassan has been given a new genetically modified skin that covers 80% of his body.

He was born with a genetic disease called junctional epidermolysis bullosa that leaves his skin as fragile as a butterfly's wings. A piece of his skin was taken, its DNA was repaired in the laboratory and the modified skin grafted back on. After nearly two years, the new skin appears completely normal.

Haemophilia (Hemophilia)

Father-of-two Jake Omer was born with haemophilia A.

Doctors say they have achieved "mind-blowing" results in an attempt to rid people of haemophilia A. The disease is caused by a genetic defect that means they do not produce a protein needed to stop bleeding.

Thirteen patients, including Jake Omer, were given the gene therapy at Barts Health NHS Trust. All are now off treatment with 11 producing near-normal levels of the protein.

Genetic Defects (BBC News 2107) Name_____ Period___
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Sickle Cell Gone

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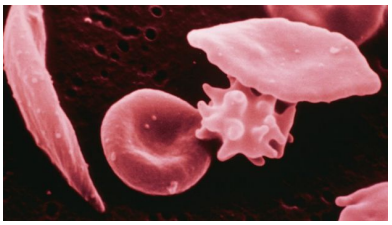

Healthy red blood cells are round, but the genetic defect makes them sickle shaped.

A French teenager's sickle cell disease was reversed using a pioneering treatment to change his DNA.

Blood cells should be round, but in the disease they become deformed and lock together to block the flow of blood around the body. Scientists altered the genetic instructions in his bone marrow so it made healthy red blood cells. So far, the therapy has worked for 15 months and the child is no longer on any medication.

The world-first procedure at Necker Children's Hospital in Paris offers hope to millions of people with the blood disorder.

Type 2 Diabetes Halved

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Isobel Murray no longer thinks of herself as diabetic.

Nearly half of patients have reversed type 2 diabetes in a "watershed" trial, say doctors in Newcastle and Glasgow. People, including Isobel Murray, spent up to five months on a low-calorie diet of soups and shakes to trigger massive weight loss.

The more weight people lost the more likely they were to put their disease into remission. The charity Diabetes UK says the trial was a landmark and had the potential to help millions of patients.

Embryos Edited

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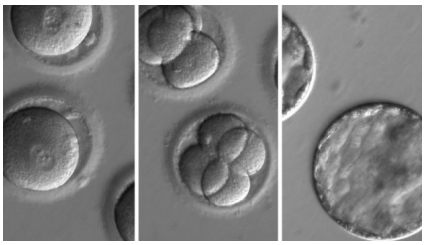

Scientists have, for the first time, successfully freed embryos of a piece of faulty DNA that causes deadly heart disease to run in families.

It potentially opens the door to preventing 10,000 disorders that are passed down the generations. The US and South Korean team allowed the embryos to develop for five days before stopping the experiment.

A gene editing technology called Crispr was used to make precise changes to the genetic code. Its applications in medicine are vast and include the idea of wiping out genetic faults that cause diseases from cystic fibrosis to breast cancer.

Note: See OregonLive Article: "OHSU Scientist Modifies DNA"