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## **EXPERT COMMENT: Gene therapies are proving their worth, but with million dollar price tags, it's not clear who should pay for them**

**Sterghios Moschos, Associate Professor in Cellular and Molecular Sciences at Northumbria University discusses the benefits of gene therapies and the financial implications of treatments.**

If you were born with a rare form of blindness, there is now a treatment for you that may restore your eyesight. That's because gene therapies became a

clinical reality in 2017. Yet many people with rare diseases that could be treated in this way may never benefit from these therapies because they are too expensive for drug companies to develop, or too costly for the patient or health service to afford. Is witnessing a starry night worth an eye-watering US\$425,000 per eye?

There are fewer things more harrowing than news that your child suffers from a rare genetic disorder that will consign them to a disabled, progressively worsening or possibly very short life. For example, spinal muscular atrophy is a debilitating, muscle-wasting disease caused by death of neurons (nerve cells) in the spine. The neurons are meant to produce a protein that is necessary for their survival, but in these patients, the levels of this protein are low to nonexistent. And the lower the level of this protein, the more the patient suffers.

The most severely affected are unable to sit and may even die before their second birthday without mechanical support for their breathing. Yet, almost a year ago, the US Food and Drug Administration approved the sale of a new drug, nusinersen, for the treatment of this disease. Nusinersen tricks the spinal neurons into using another gene to produce the protein, allowing the patient to survive.

Many doctors have rightly called the drug a miracle. There are severely affected children who received nusinersen in clinical trials and are now at school, enjoying ball games in the playground.

Hope arrived for other rare diseases, too. Genetically engineered skin stem cells restored about 80% of the skin of a seven-year-old who had suffered from blisters and open wounds from birth due to a genetic disorder. Two drug companies received approval for groundbreaking gene therapies for childhood leukemias. Another gene therapy designed for so-called bubble boy syndrome also hit the market, followed recently by a fourth for an inherited form of blindness.

Besides fixing the genomes of embryos, editing the genome of an adult has now also been attempted to fix small but devastating genetic errors.

## **Lifetime costs**

The cost of these treatments, though, ranges from about US\$500,000 to US\$1.5m. And over a lifetime, drugs like nusinersen can be even more

expensive: US\$750,000 in the first year followed by US\$375,000 a year after that – for life.

As these prices suggest, it's expensive to get a gene therapy drug to the market. It takes many years from drug design to approval. Even if the drug is approved by the regulators, costs might be so high, and patients so few, that it ultimately makes no commercial sense for drug companies to make and sell such drugs.

So far, four gene therapies have been pulled off the market, the last one being the US\$1m gene therapy, Glybera, used to treat a rare inherited disorder called lipoprotein lipase deficiency. Approved in 2012 and apparently sold to just one patient, it was eventually dropped in 2016.

## **Crowdfunding**

So, what does the future hold for gene therapy treatments and the patient's purse? Since 2015, drugs companies have started adopting pay-only-if-it-works approaches. Previous false starts illustrated the need to find new ways for patients to access specialist drugs and for payers to afford this new form of medicine. This needs a very clear definition of what "it works" means, by when and, crucially, for how long. Often, in these desperate situations, emotion can get in the way of reason, making the value of a day of life very difficult to price.

Could a crowdfunded, open-source model work? One young girl, Mila Makovec, was diagnosed with a unique mutation causing Batten's disease (a disorder of the nervous system). An American doctor, based in Boston, believes he has designed a nusinersen-like drug for Makovec that appears to be working when tested in the lab. The owners of the nusinersen technology have given the Boston doctor the freedom to use it for Makovec. Crowdfunded money has helped manufacture the drug and test it in animals, and now Makovec is being dosed with this experimental drug.

This is the apex of personalising medicine: a unique drug, probably suitable only for a single patient, ever. However, in the six months that I've been following Makovec's story, the costs have more than doubled. So far, this new drug has cost more than US\$1.6m.

## **Approving an entire class of drugs**

These successes and challenges have forced researchers, pharmaceutical companies and organisations, such as the UK's National Institute for Health and Care Excellence (NICE), to re-evaluate payment models for personalised medical treatments. One group of experts even called for approving an entire class of drugs and extrapolating between trials, drugs and possibly even diseases, helping to bring the cost down.

There is merit in this proposal; it opens the doors of hope for the handfuls of patients, or even unique patients like Makovec, that would otherwise be impossible to treat and condemned to premature, painful death. History, however, teaches us that there is no such thing as a safe family of drugs. In 2006, a phase one clinical trial for an experimental drug called TGN1412 caused an adverse reaction in the participants, all six of whom ended up in intensive care, fighting for their lives. Although TGN1412 was not a gene therapy, the story serves as a sombre reminder of things could go wrong and how experiments in cells and animals do not always prepare us for what may happen in a human.

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