

# The value of translational research in the field of rare disease

Translational research can turn promising science into new treatments for those with rare diseases. It's why we need more of it, says one expert supporting work to develop medicines.

**T**his is a hugely exciting time to be working in the area of rare diseases, says Dr Catriona Crombie, Head of the Philanthropic Fund at LifeArc, a medical research charity that helps turn promising science into real, tangible benefits for patients.

One reason for her optimism is the completion of the UK Government's 100,000 Genomes Project in 2018. This was established to sequence 100,000 genomes from around 85,000 NHS patients affected by rare diseases or cancer, with the hope of increasing understanding of genetic variants and improving diagnosis and treatment. "Breakthroughs like this give the medical community an opportunity to start to work on treatments for diseases that had previously been considered untreatable," she says.

## De-risking projects and incentivising investors

The trouble is, funds for valuable research in rare diseases are scarce, largely because of the scale of the challenge. There are around 7,000

rare disease and, at the current rate of development, it would take around 800 years to develop treatments for all rare diseases. But, developing a drug for each would provide only a small market to generate returns on investment, because of the relatively low number of patients affected per disease. A different sort of model is needed to encourage drug development, says Dr Crombie.

This is where support for translational research comes in: bridging the gap between 'bench' and 'bedside' (aka, the lab and the patient).

Financing is not the only stumbling block in rare disease treatment development. The complexity of the work requires close collaboration from a number of partners. "It would be highly unusual for one party to have the necessary skills to take a basic science concept all the way through to a treatment," explains Dr Crombie. "Translational research normally requires a number of different skillsets. It needs the biologist who understands science, the clinician who understands patient need, and experts who understand the

chemistry of the molecules under development and the experts to navigate the regulatory environment - the list is extensive. It's a multidisciplinary space, which is why we work with industry, academia, research organisations, etc."

## The need for translational research

Great Ormond Street Hospital Children's Charity (GOSH Charity) and LifeArc have a joint funding scheme focused on helping GOSH researchers drive their discoveries towards new tests and treatments for childhood rare diseases. "Researchers based at GOSH and the UCL Great Ormond Street Institute of Child Health generate potentially life-changing ideas through lab-based science," says Dr Crombie. "The hospital also has world-leading clinical expertise and, of course, a unique patient group. Through this collaboration we can bring our additional expertise in translational research, to ensure more discoveries are ready for the next stage, bringing treatments and cures one step closer

to the children that need them."

Despite her optimism that things are changing in the field, Dr Crombie stresses that this is no time for complacency. "Licensed medicines are only available for around 500 of the known rare diseases," she says. "This means that the vast number of rare disease patients receive no treatment at all. We have to do more translational research to move more potential drugs along the development pathway and ultimately provide treatments to people with challenging conditions. Otherwise some patients with rare diseases will die. It's that real... and that simple." ■

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LifeArc set up the Philanthropic Fund in 2017, which supports research with £10 million available for rare disease translational work. LifeArc's charitable model means it is able to fund in this crucial research and 'de-risk' the development of a therapeutics project. In this way, it may also make the approach attractive to potential commercial partners as an investment opportunity to take it on to the next stage of development.