



FUTURIS 4: RARE DISEASES

ENGLISH SCRIPT

02.00 Générique

02.08 Exterior of International Centre for Life, Newcastle upon Tyne (UK) and interior research laboratory

Newcastle is at the centre of a European network making significant advances in the research on inherited neuromuscular diseases. They are among rare diseases, also called "orphan" because usually orphan of research focus and market interest, as well as of public health policies.

Diseases are called rare when affect less one person per 2,000. In total, about 30 million people are affected in the European Union territory. Most of the diseases are painful, have serious consequences on life expectancy and on day-by-day life of patients and their families. And, currently, there is no cure.

02.45 ITW Louise Forrest, patient

"I think certainly as I was growing up in coming to terms with the condition, I wanted to be cured, I wanted to be told: "You do this and you can get stronger" and there's still a lot of that inside me now, even in the appointments that I had with the specialist I'm always saying "If I do this will it make me stronger?" and I'm always getting shocked, even though I know, when they say "No, it won't".

But at the same time it's important that I keep the hope, to keep doing these (physical) exercises, because I believe it's stopping me to getting weaker".

03.16 Louise Forrest in medical visit

Louise is affected by spinal muscular atrophy, a motor neuron disease, genetically transmitted, affecting muscles throughout the body, with muscles closest to the trunk most severely affected. She passed a tough surgery operation to fuse her spine and now she looks at the future with some hope.

03.41 ITW Louise Forrest

"I work full time and I would like to still continue to be more successful from that point of view. Myself and my husband would like also to have a family in the future, and hopefully the condition doesn't cause us too many problems. I do appreciate that things are going to be a lot harder for me, and my mobility could be affected and my strength will be affected again, but I hope that there is the support in place that will enable us to still go ahead and do that, and I hope continue to work for as long as possible, have a family and just lead a long and happy life".

04.19 Children at Newcastle General Hospital

Symptoms of many rare diseases appear at birth or in childhood.

The inadequate knowledge, until few years ago, of rare diseases and the scarce attention given to them by national competent authorities and the pharmaceutical industry have led to the creation of associations of patients and parents, whose actions have ensured progress in healthcare and social assistance and in raising public awareness.

Advances in diagnosing, in the past few years, permit to treat better psychological consequences and physical complication of the disorders. It's important that public health services, like here in Newcastle General Hospital, be put in place to help patients and their families to deal with the disease.

05.01

ITW PROF. KATE BUSHBY, Professor of neuromuscular genetics and Director of the muscle centre
"Children with rare neuro muscle disease can present it at any age, from tiny babies right through to teen-agers, but in most children we're beginning to pick up the signs at about the time they're starting school, when they are going to nursery and had difficulty getting up off the floor with the other children, getting difficulty keeping up with their peers at P.E. (physical education) and things like that. That's when mainly we see the first signs.

We've seen a lot of developments for the treatments of these disorders over the last few years.

We are now 20 years on from the discovery of the first gene involved in the muscle conditions and over that period people developed the animals that they can study to develop new treatments in, they shown that you can replace these faulty genes, you can upregulate some of them, you can modify them in different ways to make they function better".



05.45 intro Prof. Volker Straub, Institute of Human Genetics, University of Newcastle
About 80 per cent of rare diseases have identified genetic origins, involving one or several genes or chromosomal abnormalities. Prof. Volker Straub is leading a team of researchers on rare inherited muscular diseases.

06.00 ITW Volker Straub

"There's currently still no cure for any of the genetic muscle diseases, but I think that the encouraging news for patients is that there are a lot of concepts that came from pre-clinical studies, studies on cells, on animal models, that we're now bringing to the next step, to the clinical level.

These things will take time. If you find a new compound today, it will take probably at least 5 to maybe 10 years to get it to the market.

It's important that patients, when they get a treatment, get a safe treatment. It's important that patients know that there is evidence for the efficacy of treatment. So, I don't think that it will happen overnight".

6.53 Laboratory

After the trials on the animals, which already gave encouraging results, the research is about to start the crucial step of trials on humans. So far, important advances have been made on diagnosing genetic diseases.

07.07 ITW Debbie Hicks, Research Associate, Institute of Human Genetics, University of Newcastle

"Amongst the number of many recent advances we've made, a quite significant one was developing a better way of diagnosing patients with two very rare forms of muscular dystrophy, those caused by mistakes on the collagen 6 genes. So, from those patients, who previously gone undiagnosed by traditional methods, we've developed a method using cells from the skin, to look at the collagen 6 protein in the skin and look for abnormalities there, and that is helping us to diagnose these very rare forms of muscular dystrophy patients in a very novel way, that hasn't been tried elsewhere, and really is a massive benefit for the patients".

07.47 Laboratory

To accelerate the progress of the development of curative treatments for patients with neuromuscular diseases, a European network of excellence in research, called TREAT-NMD, is coordinated by Professor Straub together with Professor Kate Bushby.

08.07 ITW Volker Straub

"What becomes quite evident when you work with rare diseases is that there's not a single centre that has enough experience and enough patients to do clinical trials and this is what a lot of patients are waiting for: new treatments, new therapies.

So, this network of excellence really serves to develop and implement an infrastructure so that scientist and clinicians, but also industry and patients organizations are working together.

One of the problems we've noticed was that there weren't any databases that showed us how many patients there are with these specific muscle diseases. Since we started about a year ago with our network, we've now established databases throughout Europe to basically collect patients data so that we now have hundreds of patients that we can enroll into clinical trials as well".

09.09 Newcastle, pix of town

Effects from research on rare diseases will benefit directly patients affected and their families, feeling often isolated by the society. The benefits will be also for the entire population, looking at genetics as a new frontier for health.

Only a huge, European-wide, coordinated effort on research can give the hope to achieve the goal of a new era in which nobody can say: "There's no cure for me".

09.37 Générique

09.42 End