

Development in the Research of Rare Diseases

Voice-over 1:

By definition, a rare disease affects less than one in two thousand people. Symptoms are often picked up at birth or appear in early childhood. Because they've received so little attention from health authorities and the drug companies, sufferers, or more often their families, have created their own support groups and associations. By coming together in this way, they've been able to raise public awareness and improve patient care and, crucially, push for more research to be done into the diseases. It starts with diagnosis and that's where Newcastle General Hospital's Muscle Centre first begins to help patients and their families.

Kate Bushby (Director of the Muscle Centre):

Children with rare neuromuscular diseases can present it at any age, from tiny babies right through to teenagers, 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 they have difficulty getting up off the floor with the other children, (getting) difficulty keeping up with their peers at P.E. and things like that. That's when we mainly 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 twenty years on from the discovery of the first gene involved in muscle conditions and over that period people have developed the animals that they can study to develop new treatments in, they've shown that you can replace these faulty genes, you can up-regulate some of them, that you can modify them in different ways to make them function better.

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At the Institute of Human Genetics at the city's university, a team of researchers is working on rare, inherited muscular diseases. Some eighty per cent of rare diseases involve one or more genes or chromosomal abnormalities.

Volker Straub (Institute of Human Genetics):

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 probably take at least five to maybe ten years to get it to the market. It's important that patients, when they get a treatment, get a safe treatment.

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