Newcastle researchers develop life-saving treatment

Patients from across the country with a hereditary immune disorder are benefiting from life-saving treatment, thanks to the work of scientists at Newcastle University.

The researchers are the first in the world to identify the rare syndrome, which devastates the immune system and can lead to hereditary leukaemia and lung disease.  
  
A study of patients in Newcastle, funded by the blood cancer charity Leukaemia & Lymphoma Research, the Medical Research Council and the Wellcome Trust, identified four individuals with what the Newcastle scientists have labelled ‘DCML deficiency syndrome’.

The determining feature of the syndrome is that the patients have almost no ‘dendritic cells’ or ‘monocytes’ – vital cells of the immune system which originate in the bone marrow and help to fight infection.  
  
The research, published in the *Journal of Experimental Medicine,* is the first to describe dendritic cell deficiency in humans.

Without treatment, patients may die of lung disease, leukaemia or infections.

This can be prevented by a bone marrow stem cell transplant to correct the body’s immune system and prevent the risk of leukaemia.  
  
Stacey Sheppard, 23, from Sunderland, was among the first patients in the world to be diagnosed and treated for this syndrome.

She was rushed to Newcastle’s Freeman Hospital at the end of 2009 with breathing difficulties and was eventually diagnosed with ‘pulmonary alveolar proteinosis’, a rare lung problem caused by the deficiency syndrome.

Having spent months in hospital with cotinual lung infections, Stacey eventually became oxygen dependant and had to sleep with the aid of a ventilator.

Stacey said of her condition at the time: “I was so breathless that I could hardly move.

Simple little things became impossible, my weight dropped and I had no quality of life.”    
  
However, tests of Stacey’s bone marrow taken in July 2010 revealed that she had DCML deficiency, offering doctors a chance to tackle what could be the root cause of her symptoms.

She underwent a bone marrow transplant three months later, in an attempt to replace the missing cells of her immune system and repair her lungs.  
  
Stacey said: “I was told that the transplant was not guaranteed to be successful and that I may not survive, but if I didn’t have it I was given a life expectancy of one year.

The transplant has quite simply given me my life back - I can now walk short distances.

I still require oxygen but it’s considerably reduced and I no longer need a ventilator at night time. I’ve got a three-year-old girl so it’s been wonderful to be home with her and able to do activities with her that I’ve missed out on.

The difference is incredible.”  
  
[Professor Matthew Collin](http://www.ncl.ac.uk/icm/people/profile/matthew.collin), Stacey’s consultant and the scientist who led the research at the Institute of Cellular Medicine at Newcastle University, said: “The identification of this immune deficiency syndrome was important because it gave us the confidence to perform a bone marrow stem cell transplant and offer a real chance of a cure.

It is also important for Stacey to remove the risk of leukaemia and other cancers in the same way that bone marrow transplantation is already used to save the lives of many people affected by blood cancers.”  
  
Stacey’s father lost his sister to lymphoma when she was just 24 and died from lung problems himself at the age of 35.

His own father, Stacey’s granddad, had died of leukaemia in his 30s, as had his father’s sister.

Had DCML deficiency been recognised previously, their illnesses would have been understood and possibly prevented.  
  
Dr David Grant, Scientific Director at [Leukaemia & Lymphoma Research](http://www.beatbloodcancers.org/), said: “Identifying this hereditary syndrome will allow us to take action early on and prevent people from developing leukaemia, a blood cancer from which up to half of all patients still do not survive.”  
  
In 2010 Newcastle was named a ‘Centre of Excellence’ by the national blood cancer charity Leukaemia & Lymphoma Research, which has more than £7 million invested in 18 medical research projects in the area.   
  
*Press Release courtesy of the Leukaemia and Lymphoma Research charity*

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