

Gene therapy for haemophilia offers a potential cure

Haemophilia is a group of inherited disorders in which blood fails to clot properly, causing longer-than-normal bleeding. It is managed with weekly injections. Researchers in St James's Hospital took part in an international clinical trial to investigate gene therapy as a cure for patients with haemophilia. In the 54 patients in the trial, the blood-clotting factor rose by an average of 36%. This gives hope that a one-time intervention can be a permanent cure.

Currently patients with haemophilia receive prophylactic weekly injections to control the frequency and severity of bleeding and prevent joint damage or death. The condition may be mild, moderate or severe, depending on clotting-factor deficiency. Bleeding may come from gums, nose and wounds, but usually occurs internally into the knee, ankle or elbow.

The principal investigator in this gene therapy trial was Dr Niamh O'Connell, Consultant Haematologist in St James's Hospital, and

it took place at the Wellcome-HRB Clinical Research Facility (CRF) there. This innovative therapy delivers the gene into the body using a virus with adapted DNA (an adeno-associated viral vector). Patients with a new 'working copy' of genes that enable clotting

"I have lived my entire life with severe haemophilia, and I wanted to see what life would be like without severe haemophilia. And it has been very pleasant."

Brian O'Mahony

Irish Haemophilia Society CEO and gene therapy trial participant

no longer need weekly injections. In the trial, three Irish patients received the gene therapy intravenously into the liver, which started to produce the missing clotting factor.

Brian O'Mahony, a study participant and CEO of the Irish Haemophilia Society, explained that the standard prophylactic replacement therapy reduces bleeding but is not ideal. On the gene therapy he received in the clinical trial, he said: "This new treatment offers the possibility of a functional cure. The need to undergo replacement therapy several times a week take its toll. For me, taking part in this trial has increased my 'head space', in that I no longer have to constantly think about managing the condition."

