

There is one certain way to stop mitochondrial disease: prevent anyone who will develop it from being born. Until now, that has been the focus of research (Tom Whipple writes).

In some cases, using the technique known as “three parent babies”, you can replace the entire sequence of mitochondrial DNA at the fertilisation stage. In others when, as with Charlie Gard, the fault lies elsewhere in the genome, you can identify the genes shared by both parents that lead to faulty mitochondria, and select healthy embryos.

But once sufferers are born there is almost nothing that can be done.

Soon though, that may

not be the case. Scientists who have worked with mitochondrial diseases say that for the first time promising treatments are coming through that might at last mean some parents could be offered hope, even if they were too late for Charlie Gard.

Mitochondria are used by all cells to make energy. This is why when they go wrong it can be so devastating. “We can’t generate energy efficiently by any other process,” says Doug Turnbull, from Newcastle University, one of the UK’s major centres for research into the disease.

In the case of Charlie Gard, a mutation meant that he cannot produce an enzyme that in turn makes mitochondrial DNA, so mitochondria stop working. The

nucleoside technique, pioneered by the Columbia scientists who hoped to treat him, involves giving the body the tools to produce that mitochondrial DNA through a different



Charlie Gard: DNA fault

mechanism. Professor Turnbull says that an “immense amount of work” is going into that, and other drugs that could treat different mitochondrial conditions. At the moment none have

been trialled to see if they work — and none could in any case ever reverse brain damage already inflicted.

Even so, the field is more hopeful than ever. When Patrick Chinnery started working with patients suffering from mitochondrial disease, he says no one was looking into cures. Even five years ago, the Cambridge neurologist says, few were.

Now, he believes a momentum has built. “The world community is getting together. I think we’ll see something emerging soon.” It won’t be a cure, and it won’t help all sufferers, but he says it will be a start. “I would be very surprised if we don’t get a licensed treatment of some sort in the next five years.”