Three Minute Article for Parents

Thalassaemia: Current research may provide a cure for it in the future

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β-Thalassaemia is a serious disorder of blood which affects approximately 2000 children and adolescents in Sri Lanka[1]. It is incurable at present except in a minority who have undergone successful bone marrow transplantation. A majority of patients with β-thalassaemia require blood transfusions to be given monthly and drug treatment for life to reduce the iron overload[2]. Despite this, their lifespan is limited.

The excessive destruction of red blood cells (RBCs) inside the body is the main problem in patients with β-thalassaemia. Human RBCs have large quantities of oxygen carrying protein, haemoglobin, made up of two types of molecules known as α and β globin. In patients with β-thalassaemia, due to problems in the genes, the production of β-globin is reduced but the production of α-globin continues normally. This leads to an excessive accumulation of α-globin molecules which are toxic to RBCs resulting in destruction of these cells[3].

A series of studies that we published recently have shown promise that β-thalassaemia could be permanently cured by counteracting these deleterious effects in RBCs[4]. Firstly, by studying patients with β-thalassaemia, we showed that patients who have decreased productions of α-globin due to a problem in another gene have milder disease compared to those who have normal α-globin production[5]. Through observation of this natural phenomenon, we assumed that β-thalassaemia could be treated by decreasing the production of α-globin. Our next paper describes attempts to discover the ways to decrease the production of α-globin. We showed that the production of α-globin molecules in human RBCs is controlled by regions of DNA which are known as α-globin enhancers[6].

In the third paper we used 'genome editing', a novel treatment method used to alter genes, to destroy one of the α-globin enhancers in human cells in laboratories with the intention of decreasing α-globin[7]. As expected, we observed a reduction in α-globin in RBCs of patients with thalassaemia. Next, we inserted the genome edited human cells without α-globin enhancer to mice and showed that the RBCs without enhancers remain in the body of the mouse for longer periods without being destroyed.

In summary, these laboratory and animal studies illustrate that a 'cure' for β-thalassaemia could possibly be achieved by reducing α-globin production using genome editing. However, further safety studies are required before embarking on transplantation of genome edited cells into humans. It is still very early days in this novel initiative.

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