HAIRY CELL LEUKAEMIA

PERSONALISED MEDICINE IN HAEMATOLOGY

Tony's story

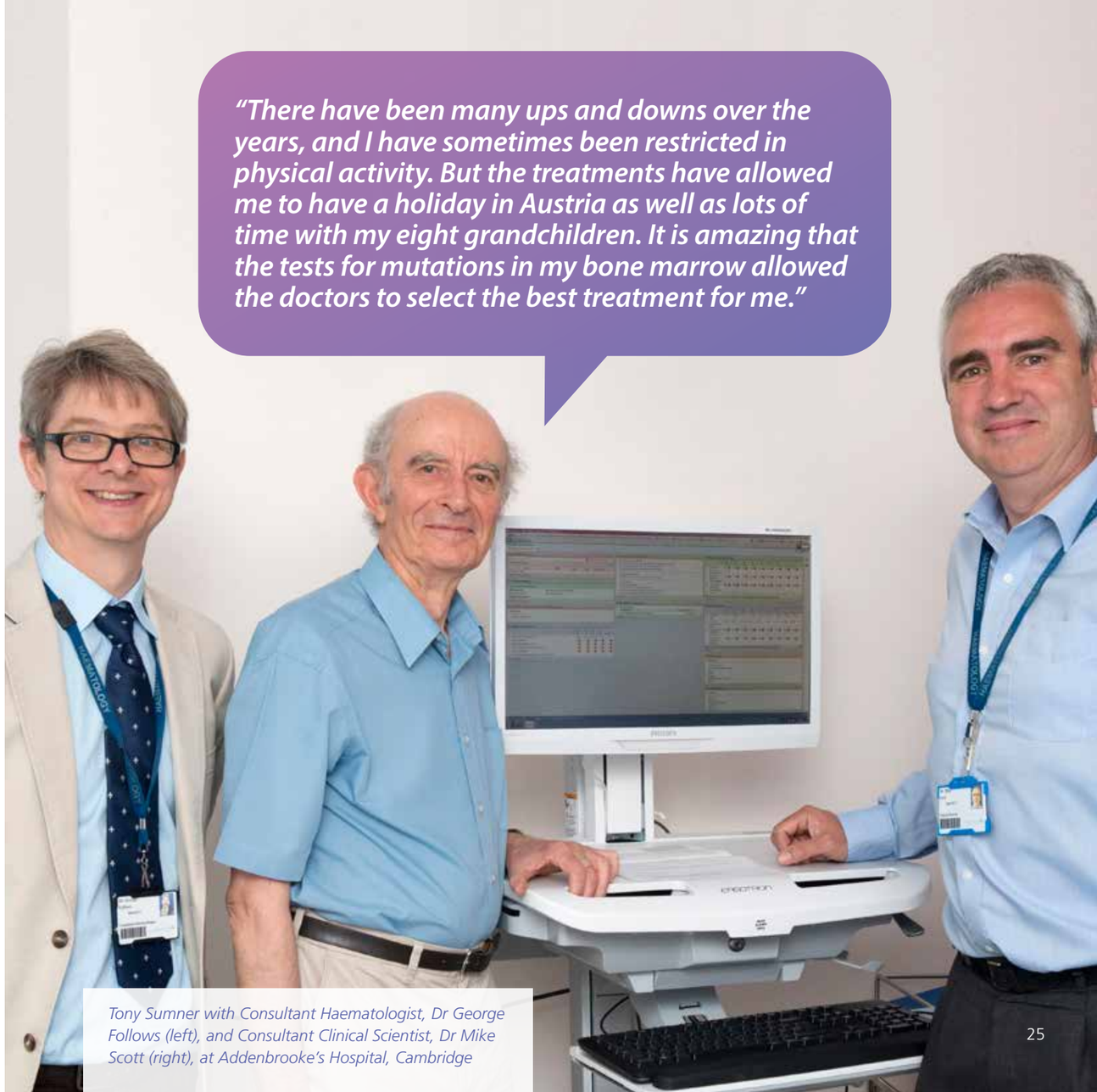
Tony Sumner's blood disorder was first diagnosed over 20 years ago when, during a routine eye test, the optician spotted a small haemorrhage at the back of the eye. He was referred to a haematology specialist, who examined samples of blood and bone marrow, and made the diagnosis of a rare form of leukaemia called hairy cell leukaemia. Tony had his enlarged spleen removed and over the next 12 years required only one block of chemotherapy treatment with a drug called Cladribine. His blood counts started to deteriorate in 2008 and further chemotherapy with Cladribine was necessary, initially supplemented by blood transfusions. In 2011, blood tests, CT scan and bone marrow samples showed the renewed presence of hairy cells, and a course of chemotherapy using Pentostatin, supported by transfusions of blood and platelets, had very limited success.

At this point, DNA analysis of the leukaemic cells showed a mutation that indicated that Tony might benefit from a new targeted therapy called Vemurafenib, usually used for the skin cancer melanoma. A low dose of this, in tablet form, has kept him remarkably well for the last four years, although recently he has had additional benefit from a drug called Rituximab.

A breakthrough using personalised medicine

Hairy cell leukaemia is a rare form of leukaemia named because of the 'hairy' appearance of the cells down the microscope. Precise diagnosis requires microscopic examination of blood and bone marrow aspirate (drawn with a needle and syringe) by a haematologist, and the review of a bone marrow trephine biopsy (small core of tissue) by a cellular pathologist. Additional techniques are used to refine the diagnosis: flow cytometry, which examines the surface profile of the cells, and molecular genetics, which defines the changes (mutations) to the DNA in the leukaemic cells. In Tony's case, there was a mutation at a specific point in the BRAF gene at position V600E. This genetic alteration is common in the skin cancer melanoma, where clinicians can now use targeted treatment with Vemurafenib for most patients with this specific mutation. Tony was only the second patient in the world to receive Vemurafenib for hairy cell leukaemia – a wonderful example of personalised medicine.

Dr George Follows
Consultant Haematologist
Cambridge University Hospitals NHS Foundation Trust



Tony Sumner with Consultant Haematologist, Dr George Follows (left), and Consultant Clinical Scientist, Dr Mike Scott (right), at Addenbrooke's Hospital, Cambridge