MEDICINE

Registry raises hope for boys like Sam

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For people such as Sam Ford, 15, who lives with the degenerative condition muscular dystrophy, the future has brightened. Sam has become the first Australian to register on a database which, experts say, significantly improves the chances of finding the right treatment for disorders like his.

The national registry of boys with Duchenne muscular dystrophy was launched in Canberra yesterday in a development which will allow Australian patients to have their cases collated with 10,000 others in the global quest for answers. About one in every 3500 Australian boys suffers DMD.

Professor Kathryn North, who heads the institute for neuroscience and muscle research at Westmead Children's Hospital, says matching patient genetic and clinical characteristics through the global database opens the way for immediate treatment trials in some cases.

"This is mind-blastingly positive," she said.

"We are immediately able to use the information [collected in the registry] to target patients with special genetically based clinical treatment."

These "genetic Band-Aids" trigger the gene to start producing the protein needed to prevent the wear and tear which ravages the muscles of dystrophy patients.

She said in an overseas case a child's condition appeared to be reversing and the child was now able to jump as a result of a targeted treatment.

The Muscular Dystrophy Foundation says the register provides a more effective link between patients, doctors and researchers, promotes a sense of community among patients and their families and better monitoring to improve care.

There are a range of genetically different causes of dystrophy diseases, each requiring a genetically specific treatment, and it is not clear whether or when the register might prove a direct benefit to Sam Ford.

But the Canberra student, whose parents say has retained a positive outlook on life, says: "I hope it will help and something will come of it."

The registry proposal was initiated by the then-parliamentary secretary for health, Jan McLucas, three years ago as a result of an appeal from a parent of a child with DMD.

Senator McLucas said the global network of registries had proven effective in improving the health and management of boys with the condition.

"I am so pleased that I can stand here today and know that child with Duchenne muscular dystrophy have increased access to life-saving treatments through clinics."

Quick off the mark … Sam Ford, the first person to register on the national DMD registry, at home in Canberra yesterday. Photo: Stefan Postles