

# Bone Marrow Failure Syndromes in Australia: improving diagnosis to inform strategies for better care and outcomes

Dr Lucy Fox

Higher Degree Fellowship (Doctoral Degree Scholarship) 2019 ongoing



Dr Lucy Fox is completing a higher degree focussing on patient outcomes in Bone Marrow Failure Syndromes (BMFS). She is the clinical research fellow at the Australian Aplastic Anaemia and other Bone Marrow Failure Syndromes Registry (AAR) and is working on analysis of the comprehensive dataset housed within the registry as part of her higher degree. She has recently launched a new AAR database and is excited to both expand the scope of the registry and continue collaborative projects, aiming to offer every Australian patient, both paediatric and adult, experiencing both acquired Aplastic Anaemia and inherited Bone Marrow Failure Syndromes the opportunity to participate in this important project. Dr Fox's additional role, as the

Bone Marrow Failure Fellow at Peter MacCallum Cancer Centre, focusses on both the inherited and acquired genetic changes that accompany the different BMFS and the way these genetic results can offer diagnostic certainty and also prognostic information to assist with clinical decision making. Her two roles described here are highly complementary and share the common primary goal of improving outcomes for all Australian patients with BMFS.

Dr Fox is intently focussed on the curation of clinical data in order to make significant contributions to clinical decision making and scientific literature. She wishes to follow and document patients over the course of



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their disease in order to learn about which patients do well and why, and why some patients do poorly. She elaborates, “We have heard from many patients of the comfort that they derive from knowing that their story is not ‘lost’, and that the particulars of their experience may one day help another family. We strive to develop our relationships with local, interstate and international colleagues so we can discuss the best testing and treatment strategies for patients, including multidisciplinary team discussion of complex cases. The importance of shared learnings and collaboration in this complex, rapidly evolving field cannot be overstated. Ultimately, we want to facilitate enrolment of Australian patients in clinical trials both locally and internationally.”

Dr Fox’s first project in this area was her involvement with the Melbourne Genomics Health Alliance Bone Marrow Failure Flagship at Peter MacCallum Cancer Centre. During this project, she met many patients with BMFS and gained an appreciation of their often long and difficult diagnostic journeys and how these diseases profoundly impact daily life. She became aware of how daunting complex ‘genetic’ conversations can be and learned how important it was to ensure improvements were instituted to the counselling and everyday support of these patients. “This is a patient group at risk of experiencing very poor outcomes, and we have much to learn in order to improve both the length and the quality of life of BMF patients. These are individually rare and diverse diseases, but share several commonalities in terms of diagnostic strategies and management. It is enormously apparent that outcomes for patients

experiencing BMFS will only be improved by collaborative efforts by researchers and clinicians around the country, and indeed the world. I am grateful for the opportunities afforded by Maddie’s Vision which have permitted me to engage with interested BMF researchers both nationally and internationally.”

Dr Fox’s commitment to the Maddie’s Vision mission is impressive, extending her professional energy beyond her core research to a myriad of other Maddie’s Vision activities. In 2019, she expertly chaired the Steering Committee of Australia’s inaugural National Symposium, was a poised master of ceremonies at the welcome event, and presented multiple times over the one and half day scientific program. In 2020, she has agreed to chair the Steering Committee for the second National Symposium (postponed till May 2021), and is chairing the subcommittee for the first ever Maddie’s Vision Patient and Family Forum, a virtual event scheduled for August 28, 2020. She is often a warm and lively face in the crowd at Maddie’s Vision fundraising events, including in 2019 the Bloody Good Dinner and Purple Ladies Lunch. Dr Fox has been instrumental in a deliberate and focussed effort to open an Australian site for the Pilot Study of Metformin in Fanconi Anaemia trial conducted at the Boston Children’s Hospital, and was the successful recipient of the opportunity to present the Maddie Riewoldt’s Vision lecture at the 10th International Congress on Shwachman Diamond Syndromes in Cambridge (now rescheduled to March 2021). Maddie Riewoldt’s Vision is delighted to be supporting her Higher Degree Fellowship.