Project Title: Characterizing a new genetic model of tibial dysplasia (twisted shin-bone)  

Host School/ Institute: The Children’s Hospital at Westmead Clinical School  
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Project Type: Laboratory based  

Project Category: Bone, Genetics, Pharmacology, Paediatrics/Child Health, Imaging, Surgery, Molecular Biology  

Project Keywords:  
1. Genetic bone disease  
2. Bone dysplasia  
3. Mouse model  
4. Neurofibromatosis  

Project Description:  
Our group has a long-standing interest in severe congenital tibial dysplasia, a condition that affects children and is a leading cause of lower limb amputation. Many of the affected children also have a genetic condition NF1 (neurofibromatosis type 1) and we have shown poor bone healing is associated with double inactivation of the NF1 gene. While we have treatments to help NF1-deficient bone healing, we are looking to prophylactically treat individuals with tibial dysplasia to prevent fractures that require multiple surgeries and/or lead to amputation.  

We are looking for an interested summer student with a background in genetics, bone biology, pathology, and/or molecular biology. The student will perform histological and molecular biology techniques on tissue samples from mice with a new virus-induced gene loss model that develops tibial bone lesions. They will also perform mechanical testing analysis on the bones. This work will give a student a range experiences with imaging, molecular, and histological techniques in a project that is highly publishable.