A New Genetic Model for Lower Urinary Tract Malformations
Friday 30th June 2017 1pm
June Lloyd Room (PUW4)

Abstract

Congenital abnormalities of the kidney and urinary tract cover a diverse spectrum, including dysplastic kidneys, hypoplastic kidneys and urinary tract obstructions. Obstructive uropathies account for around 20% of end stage renal failures in children, and even in less severe cases the obstruction still causes problems throughout life.

Treatment options for urinary tract obstructions are limited, and the genetic mechanisms behind these diseases are poorly understood. Animal models are an extremely useful tool in investigating developmental disorders, and to date few have been developed to aid in our understanding of obstructive uropathies. The T30 Homozygote (T30H) mouse was developed in the 1950s through chemical mutagenesis. This created a balanced chromosomal translocation that also resulted in an absence of smooth muscle development in the bladder. This lack of smooth muscle means that T30H mice are unable to void the bladder during gestation, which results in extensive hydronephrosis and damage to the kidneys.

It has been known for some time that the translocation was between chromosomes 2 and 11, but the exact translocation points and the genes affected were unknown. The project has three main aims, fully characterising the model both phenotypically and genetically, and then to correct the bladder smooth muscle defect in vivo.