

## **Pinpointing where kidney and lower urinary tract malformation genes are expressed in human development**

In recent years, we are beginning to appreciate that people who are born with malformed kidneys or malformed bladders can carry 'mutations' of genes.

These gene changes can sometimes run in the family, or they can occur for the first time in a specific person.

We have run NHS genetic clinics in Manchester that provide families with such genetic diagnoses, so answering their question "“why was my child born with a devastating disease?”".

This is very useful but we now need to think of ways to use this information as bases for understanding why these mutations cause the diseases.

A roadblock in this vision is that we know rather little about exactly where and when these genes are active.

In this study, we will use a very accurate and sensitive laboratory technique to pinpoint the activity of these genes as the human kidney and bladder grow before birth.

Having this information will help us understand why the growth of these organs goes wrong when the gene is mutated.

In the longer term, this information will also inform the design of futuristic treatments that make the kidney and bladder grow more normally and so prevent the need for dialysis and transplantation.