NEWS FROM THE LAB

It has been a busy year in the lab and we feel real progress has been made in understanding more about the causes of segmental overgrowth. At present we are focussing on three key questions;

1. How do genetic changes in PIK3CA and related genes cause overgrowth?
2. What is the natural history of overgrowth?
3. Are there any treatments that will help?

We remain extremely grateful for the contribution of all of our study participants and their families in our research, as without their help, none of this work would be possible.

HOW DO CHANGES IN THE GENE PIK3CA CAUSE OVERGROWTH?

In affected tissue from patients with segmental overgrowth, genetic changes (mutations) in the gene PIK3CA (also known as PI3kinase) or other related proteins lead to over-activation of the cell machinery controlling cell growth (proliferation).

WHAT CAN BE LEARNT ABOUT THE NATURAL HISTORY OF OVERGROWTH? We have learnt that some people with segmental overgrowth carry on growing, whereas some stop growing when they become a young adult. Why is this? To answer this question we have made some antibodies that will detect the mutant form of the protein encoded by the gene PIK3CA, which will allow us to identify precisely which cells and tissues are affected. The cells in the picture below are called “hybridomas” and are like small factories, which produce the antibodies. This work will start in early 2015, and we are excited about what can be learnt here, and hope it will help us to make better predictions about how growth will progress in affected patients.
ARE THERE ANY TREATMENTS THAT WILL HELP?

We have been testing a number of different treatments in the lab on cells grown from our study participants. We have found that several treatments are able to switch off growth signals in affected cells, and slow down growth. One of these drugs is called sirolimus (also known as rapamycin), and is currently used to treat patients with kidney transplants. From this work we think this is the first drug that should be tested in clinical trials.

UPDATE ON CLINICAL TRIALS

The design of a clinical trial of sirolimus treatment has been finalised and the plans are currently under review by research experts and ethical committees. We anticipate the trial will start in spring 2015 and is for anyone with a confirmed genetic change in PIK3CA and progressive overgrowth. The trial will involve receiving sirolimus treatment for six months with scans before and after treatment to test if the drug works. If you are interested in the clinical trial please get in touch.

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