OLLIER DISEASE & MAFFUCCI SYNDROME
Research into Ollier Disease and Maffucci Syndrome and cartilaginous tumours (chondrosarcoma).

Ollier disease/ Maffucci syndrome is a rare ‘orphan’ disease affecting less than 100,000 of the population. These syndromes are characterised by multiple cartilaginous tumours, and vascular tumours – heamangiomas- in the case of Maffucci syndrome.

Since we started our research on chondrosarcoma, we have shown that about 50% of patients with the disease have a genetic alteration known as IDH1 in their tumour. Preliminary data from a pilot study shows that we can detect this alteration in the blood and is potentially useful for monitoring patients for recurrence of the tumour.

We need your help for further research to show that this new test can be incorporated into the NHS routine testing.

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