Lay Summary

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Verifying the surrogate marker, NCOA2, for identifying the HEY1-NCOA2 fusion gene in mesenchymal chondrosarcoma (MCS)

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Mesenchymal chondrosarcoma (MCS) is a rare cancer of the bone and cartilage affecting young people. It is highly aggressive and often fatal. MCS is difficult to diagnose and often patients are given the incorrect diagnosis. This is because the diagnosis depends on identifying a specific genetic abnormality in the tumour, called a HEY1-NCOA2 gene fusion, which is technically challenging to detect. This study aims to make the diagnosis of MCS faster and therefore promote earlier detection and treatment. One way of identifying the gene fusion is a “surrogate antibody” which is simple technique to perform and can indirectly indicate the presence of the fusion gene. Surrogate antibodies are widely used in other tumours defined by fusion genes. To date, no study has validated any potential surrogate markers for MCS. We propose to test a candidate surrogate marker, an NCOA2 antibody. If successful, this will greatly benefit patients and further research for this often fatal disease.