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Information about the contributions of each person named as having participated in the study

- 1) Guarantor(s), i.e., person(s) who is (are) responsible for the integrity of the work as a whole:
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- 2) Authors who participated in the conception of the study: Chris Bacon, Nick Bown
- 3) Design & Methods. The following authors were responsible for specific investigations (please
- Clare Bedwell & Nick Bown were responsible for design, acquisition, analysis & interpretation of cytogenetic data.
- David Rowe & Nick Bown were responsible for design, acquisition, analysis and interpretation of molecular genetic data.
- Deborah Moulton & Gail Jones were responsible for acquisition, analysis and interpretation of clinical data.
- Chris Bacon was responsible for design, acquisition, analysis & interpretation of pathology data and for interpretation of cytogenetic and molecular genetic data
- 4) **Results**. The following authors were responsible for specific portions of the results, including figures and tables (please indicate the person responsible for each figure and each table):
- Chris Bacon was responsible for data in Fig 1A & B
- Clare Bedwell & Nick Bown were responsible for data in Fig 1C, D, E & F
- David Rowe & Nick Bown were responsible for data in Fig 1G
- 5) Writing the manuscript. The following authors were responsible for writing the manuscript:
- Chris Bacon was primarily responsible for writing the manuscript
- All authors contributed to the review and revision of the manuscript for technical and intellectual content
- All authors approved the final version of the manuscript for publication
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- Lisa Grady (Northern Genetics Service) DNA sequencing