

The Victorian Cancer Cytogenetics Service, in addition to providing a statewide service for cancer cytogenetics, has a research interest in the underlying genetic events that arise from chromosome rearrangements. We seek to determine how these rearrangements cause haematological malignancies, particularly acute myeloid leukaemia and myelodysplastic syndromes.

### About us

The Victorian Cancer Cytogenetics Service (VCCS) receives more than 4,500 specimens annually for cytogenetic analysis from all over Victoria and performs in excess of 1,000 fluorescence in situ hybridisation (FISH) tests per year.

Chromosome analysis has become an integral part of the evaluation of patients with haematological malignancies. The VCCS receives approximately 200 new cases of acute myeloid leukaemia, 70 to 80 new cases of chronic myeloid leukaemia and 50 new cases of acute lymphoblastic leukaemia annually for chromosome analysis, plus many review cases and other more chronic

disorders such as myelodysplastic syndromes, myeloproliferative disorders, lymphomas, myelomas and some non-haematological disorders such as Ewing tumours and other sarcomas. The chromosome abnormalities identified in these malignancies provide diagnostic and prognostic information to aid clinical management. FISH is also used to establish the presence of chromosome abnormalities in non-dividing cells and to elucidate complex abnormalities.

The research activities of the VCCS include developmental activities designed to improve methods of providing cancer cytogenetic analyses, reporting of novel chromosome abnormalities or series of cases that have been identified in the course of the clinical work of the department; and a study of acute myeloid leukaemia (AML) and myelodysplastic syndrome (MDS) cases with deletions of the long arm of one chromosome – 20-del(20q) – to identify the underlying genes that are disregulated in this common chromosome abnormality.

### Projects in progress or completed

Ongoing studies by Dr Ruth MacKinnon (complex chromosome abnormalities in cases of acute myeloid leukaemia and myelodysplastic syndromes) have led to the development of carcinogenesis in these patients. We propose that loss of part of the long arm of chromosome 20 (20q) not only knocks out a tumour suppressor but

is also accompanied by amplification of an adjacent part of 20q containing an oncogene which contributes to the development of AML/MDS in these patients. This oncogene may be deregulated by the loss of the tumour suppressor gene in the adjacent part of the chromosome and we believe that this model may apply to other regions commonly deleted in AML/MDS. We have received three-year funding from the Cancer Council of Victoria to employ Patricia Susanto as a research assistant to further investigate potential oncogenes on 20q, starting from January 2009.

### The team

**Assoc Prof Lynda Campbell, Medical Director**; Dr Ruth MacKinnon, Senior Research Officer; Patricia Susanto, Research Assistant

### Highlights

Dr Ruth MacKinnon, Second Prize, Best Poster (Scientific), St Vincent's Research Week, 2008

### Grants

*Nandurkar H, Campbell L, Wei A, MacKinnon R*

Identification of new oncogenes in the development of acute myeloid leukaemias and myelodysplastic syndromes. Cancer Council of Victoria, (2008), \$62,750

## Selected presentations

### *Campbell L*

- Invited speaker, '*Cytogenetics and FISH in Myeloid Disorders*', International Society of Laboratory Haematology, Sydney, May 2008
- Invited speaker, '*Art and science informing haematology*', Carl de Grouchy Oration – St. Vincent's, Melbourne, September 2008
- Invited speaker, '*Cytogenetic analysis of acute leukaemias*', Australian Institute of Medical Scientists National Scientific Meeting, Melbourne, October 2008
- Invited speaker, '*Unravelling the complexity of chromosome abnormalities in acute myeloid leukaemia*', Haematology Society of Australia and New Zealand Annual Scientific Meeting, Perth, October 2008

## Publications

Henderson MJ, Choi S, Beesley AH, Baker DL, Wright D, Papa RA, Murch A, Campbell LJ, Lock RB, Norris MD, Haber M, Kees UR 2008 'A xenograft model of infant leukaemia reveals a complex MLL translocation', *British Journal of Haematology*, 140, 716-719