

MEETING ABSTRACT

Open Access

Dissecting genetic pathways in schwannomatosis and malignant rhabdoid tumour

E Algar

From Familial Aspects of Cancer 2011 Research and Practice: A combined meeting of kConFab, Australian Breast Cancer Family Study, Australian Colorectal Cancer Family Study, Australian Ovarian Cancer Study, Family Cancer Clinics of Australia and New Zealand and kConFab Kingscliff, Australia. 23-26 August 2011

Schwannomatosis is a form of Neurofibromatosis type 2 (NF2) characterized by multiple schwannomas without vestibular involvement, affecting the cranium, spine and periphery. Several recent genetic studies have implicated the *SMARCB1/INI1* tumour suppressor gene in familial schwannomatosis. *SMARCB1* is located centromeric to *NF2* on 22q and loss of function of *SMARCB1* is also a hallmark of malignant rhabdoid tumour (MRT), a highly aggressive tumour of infancy. Both familial and sporadic schwannoma tumours show a mosaic pattern of *SMARCB1* protein expression, suggestive of tumour cells either haploinsufficient, or null for *SMARCB1* protein. Familial schwannomas linked to constitutional *SMARCB1* mutation can also have somatic mutation of *NF2*, and conversely, schwannoma tumours associated with constitutional *NF2* mutation show mosaic loss of *SMARCB1*, suggesting the involvement of a four-hit mechanism. Molecular analysis for evidence of constitutional *SMARCB1* mutation is important in both familial and sporadically occurring schwannomatosis because of the transmission risk for a mutation predisposing to the incurable MRT, in early childhood. However as for *NF2*, recent evidence suggests that constitutional *SMARCB1* mutations have variable penetrance and exhibit mosaicism, highlighting the importance of examining multiple tumour tissue samples as well as blood in affected individuals to ascertain germline predisposition and to provide accurate counseling for transmission risk. Further studies are needed to define the *SMARCB1* mutation spectrum in schwannomatosis and to dissect the strikingly different biology between schwannoma and MRT.

Published: 12 April 2012

doi:10.1186/1897-4287-10-S2-A8

Cite this article as: Algar: Dissecting genetic pathways in schwannomatosis and malignant rhabdoid tumour. *Hereditary Cancer in Clinical Practice* 2012 **10**(Suppl 2):A8.

Submit your next manuscript to BioMed Central
and take full advantage of:

- Convenient online submission
- Thorough peer review
- No space constraints or color figure charges
- Immediate publication on acceptance
- Inclusion in PubMed, CAS, Scopus and Google Scholar
- Research which is freely available for redistribution

Submit your manuscript at
www.biomedcentral.com/submit



Correspondence: elizabeth.algar@rch.org.au
Molecular Oncology Laboratory, Murdoch Children's Research Institute, Royal Children's Hospital, Flemington Rd, Parkville, 3052, Australia



© 2012 Algar; licensee BioMed Central Ltd. This is an Open Access article distributed under the terms of the Creative Commons Attribution License (<http://creativecommons.org/licenses/by/2.0>), which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.