

## William Foulkes-Round 17

### Mutations in DICER1 and related microRNA processing genes in dysplasia, dystonogenic diseases, tumours and other conditions

*Funded in partnership with Childhood Cancer Canada Foundation.*

#### **Publications:**

1. de Kock L, Bah I, Revil T, Bérubé P, Wu MW, Sabbaghian N, Priest JR, Ragoussis J, Foulkes WD. Deep Sequencing Reveals Spatially Distributed Distinct Hot Spot Mutations in DICER1-Related Multinodular Goiter. *J Clin Endocrinol Metab* 2016 Oct;101(10):3637-3645. Epub 2016 Jul 26
2. de Kock L, Terzic T, McCluggage WG, Stewart CJR, Shaw P, Foulkes WD, Clarke BA. DICER1 Mutations are Consistently Present in Moderately and Poorly Differentiated Sertoli-Leydig Cell Tumours. *Am J Surg Pathol*. 2017 Sep;41(9):1178-1187. doi: 10.1097/PAS.0000000000000895
3. de Kock L, Rivera B, Revil T, Thorner P, Goudie C, Bouron-Dal Soglio D, Choong CS, Priest JR, van Diest PJ, Tanboon J, Wagner A, Ragoussis J, Choong PF, Foulkes WD. Sequencing of DICER1 in sarcomas identifies biallelic somatic DICER1 mutations in an adult-onset embryonal rhabdomyosarcoma. *Br J Cancer*. 2017 Jun 6;116(12):1621-1626. doi: 10.1038/bjc.2017.147. Epub 2017 May 18.
4. Saskin A, de Kock L, Sabbaghian N, Apellaniz-Ruiz M, Bozkurt C, Bouron-Dal Soglio D, Foulkes WD. A case of neuroblastoma in DICER1 syndrome: Chance finding or noncanonical causation? *Pediatr Blood Cancer*. 2017 Aug 2. doi: 10.1002/pbc.26715. [Epub ahead of print]

#### **Invited Presentations:**

1. (September 2016). From Common to Rare: Travels in Ovarian Cancer. Oncology Grand Rounds, UNC Lineberger Comprehensive Cancer Center, Chapel Hill, United States
2. (October 2016). New genes for rare diseases DICER1 and SMARCA4 – what they do, what they cause? The Bellvitge Institute for Biomedical Research (IDIBELL), Barcelona, Spain
3. (November 2016). DICER1: mutations and phenotypes Canadian Association of Genetic Counsellors. Omni Hotel, Montreal, Quebec
4. (March 2017). New neuroendocrine tumour syndromes. University of Calgary, Calgary, Canada
5. (May 2017). DICER1 in health and disease. Poznan University, Poznan, Poland
6. (May 2016) DICER1 and endocrine tumors. ECE 2016: S24 - Symposium 24: Endocrine neoplasias: new associations Room: SAAL 13B, May 31, 2016, Munich, Germany

#### **Conference and Poster Presentations:**

1. de Kock et al. Detection of somatic mosaicism in children with suspected DICER1 syndrome using high sensitivity sequencing with molecular tag-containing Haloplex HS. (Abstract/Program #2715). Presented at the Annual Meeting of The American Society of Human Genetics, October 7, 2015 in Baltimore, MD.