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IN BRIEF

THYROID CANCER

Occult disease risks quantified

Patients with thyroid cancer who undergo thyroidectomy are at risk of harbouring occult disease, owing to undetected tumour invasion of the lymph nodes; however, the extent of lymph-node examination required to determine this risk is unknown. Now, findings from a National Cancer Database study of 78,724 patients reveal that 53% of patients who were considered not to have occult disease, on the basis of examination of only a single lymph node, were falsely identified as 'node-negative'. Further analyses indicate that in order to rule out occult nodal disease with a 90% level of certainty, clinicians should examine six, nine and 18 nodes in patients undergoing thyroidectomy with stages T1b, T2, or T3 disease, respectively. These data provide the best available evidence to date on the extent of lymph-node inspection required to rule out the possibilty of occult disease in patients with thyroid cancer, and will hopefully result in better prognostification of patients following thyroidectomy.

ORIGINAL ARTICLE Robinson, T. J. *et al.* How many lymph nodes are enough? Assessing the adequacy of lymph node yield for papillary thyroid cancer. *J. Clin. Oncol.* http://dx.doi.org/10.1200/JCO.2016.67.6437 (2016).

GENETICS

Novel oesophageal cancer risk loci identified

Barrett's oesophagus, a condition usually caused by chronic exposure to stomach acid, is a precursor state to the development of oesophageal adenocarcinoma; however, only a small fraction of those with Barrett's oesophagus develop cancer. Data from a newly published meta-analysis of genome-wide association studies of this association reveal the presence of 14 genetic risk loci that confer an increased risk of both Barrett's oesophagus and oesophageal adenocarcinoma. A number of these genetic risk loci were previously unreported, meaning that they were not detected in the cohorts of each individual study alone. The strongest association among any of the newly identified risk loci was with a single-nucleotide polymorphism in the *CFTR* gene. This association is supported by the established link between cystic fibrosis and oesophageal cancer; although, the mechanisms of both this link, and the association reported following this analysis are unclear.

ORIGINAL ARTICLE Gharahkhani, P. *et al.* Genome-wide association studies in oesophageal adenocarcinoma and Barrett's oesophagus: a large-scale meta-analysis. *Lancet Oncol.* http://dx.doi.org/10.1016/S1470-2045(16)30240-6 (2016).

HAEMATOLOGICAL CANCER

Shelterin complex mutated in familial CLL

The existence of a familal risk of chronic lymphocytic leukaemia (CLL) has been established for some time, although, thus far, evidence of germ-line alleles that might explain this risk has been lacking. Now, researchers have shown that loss-of-function variations in *POT1*, a shelterin complex protein involved in telomere maintenance, co-segregated with CLL in four families. Co-segregation of mutations in *ACD* and *TERF2IP*, which encode other components of the shelterin complex was observed in a further three families. Further investigations revealed that having the p.Gln376Arg variant of *POT1* confers a 3.61-fold increase in the risk of CLL in an analysis of 1,083 patients with CLL and 5,854 individuals without the disease. These findings demonstrate the existence of germ-line variants that might explain certain aspects of the familial risk of CLL.

ORIGINAL ARTICLE Speedy, H. E. *et al.* Germline mutations in shelterin complex genes are associated with familial chronic lymphocytic leukemia. *Blood* http://dx.doi.org/10.1182/blood-2016-01-695692 (2016).