IN BRIEF

BREAST CANCER

Single nucleotide polymorphisms associated with breast cancer risk in *BRCA1* mutation carriers

Antoniou, A. C. *et al. Nat. Genet.* **42**, 885–892 (2010)

BRCA mutations confer a high risk of developing breast and ovarian cancer, and genetic variants may modify this risk. In a genome-wide association study, 2,383 *BRCA1* carriers with and without breast cancer were genotyped, and 96 single-nucleotide polymorphisms (SNPs) were identified from the comparison between the two groups. In the second stage of the study, 89 SNPs were examined in a further 5,986 *BRCA1* carriers. On 19p13, five SNPs were related to risk of breast cancer, and also associated with estrogen-receptor-negative and triple-negative breast cancer. Uncovering genetic variation may be useful in individual cancer risk assessment.

BIOMARKERS

Measuring thyrotropin receptor mRNA levels as a novel biomarker to assess thyroid cancer status

Milas, M. *et al. Ann. Surg.* **252**, 643–651 (2010)

A study published in the *Annals of Surgery* demonstrates the utility of a novel marker to assess disease status in patients with thyroid cancer. Levels of thyrotropin receptor mRNA were measured in blood from patients undergoing thyroid surgery, postoperatively, and in patients with known benign disease. Levels of mRNA had a 96% predictive value for differentiated thyroid cancer, but mRNA levels became undetectable on the first day after surgery in all patients except those who developed cervical disease within 1 year. Thyrotropin receptor mRNA was also undetectable in patients with benign disease. This biomarker represents an additional tool for the evaluation of patients with thyroid disease.

HEMATOLOGY

Malignant stem cells contribute to treatment failure in patients with 5q deletion myelodysplastic syndrome

Tehranchi, R. *et al. N. Engl. J. Med.* **363**, 1025–1037 (2010)

Patients with 5q deletion (del[5q]) myelodysplasia respond well to lenalidomide, and complete remission is frequently observed. However, 50% of patients have relapse after 2–3 years. To address whether malignant stem cells are responsible for disease progression, a study analyzed stem cells and progenitor cells from seven patients who had become transfusion independent, and one control patient who did not respond to lenalidomide. Before treatment, almost all $CD34^+$,$CD38^+$ progenitors and $CD34^+$,$CD38^-$/low, $CD90^+$ stem cells had the 5q deletion, but these cells were reduced in patients in remission. Resistance to treatment, however, was associated with recurrence or expansion of the del(5q) clones, leading to clinical progression.