DNA Sequencing Publications

Standard Sequencing

1. Carro MS et al.
   *DEK Expression is controlled by E2F and deregulated in diverse tumor types.*
   Cell Cycle. 2006 Jun;5(11)

2. Lassandro L et al.
   *The DNA sequence quality machine at IFOM: a simple Web-based tool for quantitative assessment of sequencing reactions.*

Full exon sequencing

1. Salsano E et al.
   *An autoimmune neurological disease due to interleukin 6 hypersecretion.*
   J Neuroinflammation. 2013 Feb 21;10:29

   *The lymphoma-associated NPM-ALK oncogene elicits a p16INK4a/pRb-dependent tumor-suppressive pathway.*

3. Westhoff B et al.
   *Alterations of the Notch pathway in lung cancer.*
   Proc Natl Acad Sci U S A. 2009 Dec 29;106(52):22293-8

Mutation, SNP, Variant Analysis

1. De Grassi A et al.
   *Deep sequencing of the X chromosome reveals the proliferation history of colorectal adenomas.*

2. Belloni E et al.
   *Genomic characterization of asymptomatic CT-detected lung cancers.*
   Genomic characterization of asymptomatic CT-detected lung cancers.

3. De Grassi A et al.
   *Ultradeep sequencing of a human ultraconserved region reveals somatic and constitutional genomic instability.*
   PLoS Biol. 2010 Jan;8(1)
4 Falini B et al.
**Immunohistochemistry predicts nucleophosmin (NPM) mutations in acute myeloid leukemia.**

5 Colombo E et al.
**Delocalization and destabilization of the Arf tumor suppressor by the leukemia-associated NPM mutant.**

6 Mariano AR et al.
**Cytoplasmic localization of NPM in myeloid leukemias is dictated by gain-of-function mutations that create a functional nuclear export signal.**

7 Alcalay M et al.
**Acute myeloid leukemia bearing cytoplasmic nucleophosmin (NPMc+ AML) shows a distinct gene expression profile characterized by up-regulation of genes involved in stem-cell maintenance.**
Blood. 2005 Aug 1;106(3):1999-2005

Clinical (in collaboration with CGT Lab)

1 Catucci I et al.
**Haplotype analyses of the c.1027C>T and c.2167_2168delAT recurrent truncating mutations in the breast cancer-predisposing gene PALB2.**

2 Ovarian Cancer Association Consortium et al.
**No clinical utility of KRAS variant rs61764370 for ovarian or breast cancer.**
Gynecol Oncol. 2016 May;141(2):386-401

3 Pirie A et al.
**Common germline polymorphisms associated with breast cancer-specific survival.**
Breast Cancer Res. 2015 Apr 22;17:58

4 Kabisch M et al.
**Inherited variants in the inner centromere protein (INCENP) gene of the chromosomal passenger complex contribute to the susceptibility of ER-negative breast cancer.**
Carcinogenesis. 2015 Feb;36(2):256-71
5 Kuchenbaecker KB et al.  
*Identification of six new susceptibility loci for invasive epithelial ovarian cancer.*  
Nat Genet. 2015 Feb;47(2):164-71

6 Ghoussaini M et al.  
*Evidence that breast cancer risk at the 2q35 locus is mediated through IGFBP5 regulation.*  

7 Sawyer E et al.  
*Genetic predisposition to in situ and invasive lobular carcinoma of the breast.*  

8 Osorio A et al.  
*DNA glycosylases involved in base excision repair may be associated with cancer risk in BRCA1 and BRCA2 mutation carriers.*  

9 Peterlongo P et al.  
*Candidate genetic modifiers for breast and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers.*  
Cancer Epidemiol Biomarkers Prev. 2015 Jan;24(1):308-16

10 Couch FJ et al.  
*Genome-wide association study in BRCA1 mutation carriers identifies novel loci associated with breast and ovarian cancer risk.*  

11 Bojesen SE et al.  
*Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer.*  

12 Colombo M et al.  
*Comparative in vitro and in silico analyses of variants in splicing regions of BRCA1 and BRCA2 genes and characterization of novel pathogenic mutations.*  

13 Brewster BL et al.  
*Identification of fifteen novel germline variants in the BRCA1 3’UTR reveals a variant in a breast cancer case that introduces a functional miR-103 target site.*  
Hum Mutat. 2012 Dec;33(12):1665-75.
14. Catucci I et al.
*Germline mutations in BRIP1 and PALB2 in Jewish high cancer risk families.*
Fam Cancer. 2012 Jun 13

15. Mavaddat N et al.
*Pathology of breast and ovarian cancers among BRCA1 and BRCA2 mutation carriers: results from the Consortium of Investigators of Modifiers of BRCA1/2 (CIMBA).*

16. Catucci I et al.
*The CASP8 rs3834129 polymorphism and breast cancer risk in BRCA1 mutation carriers.*

17. De Vecchi G et al.
*The p53 Arg72Pro and Ins16bp polymorphisms and their haplotypes are not associated with breast cancer risk in BRCA-mutation negative familial cases.*

18. Catucci I et al.
*PALB2 sequencing in Italian familial breast cancer cases reveals a high-risk mutation recurrent in the province of Bergamo.*
Genet Med. 2014 Sep;16(9):688-94.

19. French D et al.
*Functional variants at the 11q13 risk locus for breast cancer regulate cyclin D1 expression through long-range enhancers.*

20. Catucci I et al.
*The SNP rs895819 in miR-27a is not associated with familial breast cancer risk in Italians.*

21. Peterlongo P et al
*Candidate genetic modifiers for breast and ovarian cancer risk in BRCA1 and BRCA2 mutation carriers.*

22. Purrington KS et al.
*Genetic variation in mitotic regulatory pathway genes is associated with breast tumor grade.*
Bojesen SE et al.  
*Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer.*

Colombo M et al.  
*Comparative in vitro and in silico analyses of variants in splicing regions of BRCA1 and BRCA2 genes and characterization of novel pathogenic mutations.*

Catucci I et al.  
*Sequencing analysis of SLX4/FANCP gene in Italian familial breast cancer cases.*

Peterlongo P et al.  
*The rs1297533 variant in the miR-125a and breast cancer risk in Germany, Italy, Australia and Spain.*

Verderio P et al.  
*A BRCA1 promoter variant (rs11655505) and breast cancer risk.*

Catucci I et al.  
*Evaluation of SNPs in miR-146a, miR196a2 and miR-499 as low-penetrance alleles in German and Italian familial breast cancer cases.*

**NGS data validation**

1 Belloni E et al.  
*Whole-exome sequencing identifies driver mutations in asymptomatic CT-detected lung cancers with normal karyotype.*
Accettato in Febbraio 2015

2 Riva L et al.  
*Acute promyelocytic leukemias share cooperative mutations with other myeloid-leukemia subgroups.*
Blood Cancer J. 2014 Mar 21

3 Bodini M et al.  
*The hidden genomic landscape of acute myeloid leukemia: subclonal structure revealed by undetected mutations.*
Blood. 2014 Dec 12.