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)

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2 Lassandro L et al.

The DNA sequence quality machine at IFOM: a simple Web-based tool for quantitative assessment of sequencing reactions.

DNA Seq. 2003 Aug;14(4):327-30.

Full exon sequencing

1 Salsano E et al.

An autoinflammatory neurological disease due to interleukin 6 hypersecretion.

J Neuroinflammation. 2013 Feb 21;10:29)

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2 Martinelli P et al.

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

Blood. 2011 Jun 16;117(24):6617-26.

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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.

Genome Biol. 2014 Aug 30;15(8):437.

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2 Belloni E et al.

Genomic characterization of asymptomatic CT-detected lung cancers.

Genomic characterization of asymptomatic CT-detected lung cancers.

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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.
Blood. 2006 Sep 15;108(6):1999-2005

5 Colombo E et al.
Delocalization and destabilization of the Arf tumor suppressor by the leukemia-associated NPM mutant.
Cancer Res. 2006 Mar 15;66(6):3044-50.

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.
Oncogene. 2006 Jul 20;25(31):4376-80.

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):

8 Dell'Agnola C et al.
In vitro and in vivo hematopoietic potential of human stem cells residing in muscle tissue.
Exp Hematol. 2002 Aug;30(8):905-14.

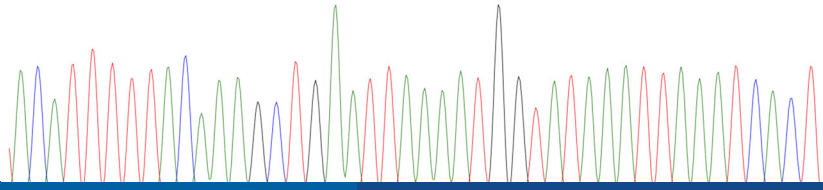
Clinical (in collaboration with CGT Lab)

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.
Breast Cancer Res Treat. 2016 Nov;160(1):121-129

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.
Nat Commun. 2014 Sep 23;4:4999.

7 Sawyer E et al.
Genetic predisposition to in situ and invasive lobular carcinoma of the breast.
PLoS Genet. 2014 Apr 17;10(4):e1004285.

8 Osorio A et al.
DNA glycosylases involved in base excision repair may be associated with cancer risk in BRCA1 and BRCA2 mutation carriers.
PLoS Genet. 2014 Apr 3;10(4):e1004256.

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.
PLoS Genet. 2013;9(3):e1003212.

11 Bojesen SE et al.
Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer.
Nat Genet. 2013 Apr;45(4):371-84, 384e1-2

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.
PLoS One 2013; 8(2):e57173

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).
Cancer Epidemiol Biomarkers Prev. 2012 Jan;21(1):134-47

16 Catucci I et al.
The CASP8 rs3834129 polymorphism and breast cancer risk in BRCA1 mutation carriers.
Breast Cancer Res Treat. 2011 Feb;125(3):855-60.

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.
Cancer Detect Prev. 2008;32(2):140-3.

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.
Am J Hum Genet 2013 Apr 27;92(4):489-503.

20 Catucci I et al.
The SNP rs895819 in miR-27a is not associated with familial breast cancer risk in Italians.
Breast Cancer Res Treat. 2012 Jun;133(2):805-7.

21 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.

22 Purrington KS et al.
Genetic variation in mitotic regulatory pathway genes is associated with breast tumor grade.
Hum Mol Genet. 2014 Nov 15;23(22):6034-46.

23 Bojesen SE et al.
Multiple independent variants at the TERT locus are associated with telomere length and risks of breast and ovarian cancer.
Nat Genet. 2013 Apr;45(4):371-84, 384e1-2.

24 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.
PLoS One. 2013;8(2):e57173.

25 Catucci I et al.
Sequencing analysis of SLX4/FANCP gene in Italian familial breast cancer cases.
PLoS One. 2012;7(2):e31038.

26 Peterlongo P et al.
The rs12975333 variant in the miR-125a and breast cancer risk in Germany, Italy, Australia and Spain.
J Med Genet. 2011 Oct;48(10):703-4.

27 Verderio P et al.
A BRCA1 promoter variant (rs11655505) and breast cancer risk.
J Med Genet. 2010 Apr;47(4):268-70.

28 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.
Hum Mutat. 2010 Jan;31(1):E1052-7.

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.

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