

Selected Publications Citing **SURVEYOR® Nuclease** for Mutation Detection and Analysis

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Oncology

DHPLC/SURVEYOR Nuclease: A Sensitive, Rapid and Affordable Method to Analyze BRCA1 and BRCA2 Mutations in Breast Cancer Families.

Pilato B, De Summa S, Danza K, Papadimitriou S, Zaccagna P, Paradiso A, Tommasi S.
Mol Biotechnol. Nov 10 (2011). [Epub ahead of print]

Phosphatidylinositol-3-kinase alpha catalytic subunit mutation and response to neoadjuvant endocrine therapy for estrogen receptor positive breast cancer.

Ellis MJ, Lin L, Crowder R, Tao Y, Hoog J, Snider J, Davies S, DeSchryver K, Evans DB, Steinseifer J, Bandaru R, Liu W, Gardner H, Semiglazov V, Watson M, Hunt K, Olson J, Baselga J.
Breast Cancer Res. Treat. 119, 379-390 (2010).

Detection of androgen receptor mutations in circulating tumor cells in castration-resistant prostate cancer.

Jiang Y, Palma JF, Agus DB, Wang Y, Gross ME.
Clin. Chem. 58, 1492-1495 (2010).

Associations between polycyclic aromatic hydrocarbon-related exposures and p53 mutations in breast tumors.

Mordukhovich I, Rossner P Jr, Terry MB, Santella R, Zhang YJ, Hibshoosh H, Memeo L, Mansukhani M, Long CM, Garbowski G, Agrawal M, Gaudet MM, Steck SE, Sagiv SK, Eng SM, Teitelbaum SL, Neugut AI, Conway-Dorsey K, Gammon MD.
Environ Health Perspect. 118, 511-8 (2010).

Noninvasive detection of EGFR T790M in gefitinib or erlotinib resistant non-small cell lung cancer.

Kuang Y, Rogers A, Yeap BY, Wang L, Makrigiorgos M, Vetrand K, Thiede S, Distel RJ, Jänne PA.
Clin Cancer Res. 15, 2630-6 (2009).

Strain-specific spontaneous and NNK-mediated tumorigenesis in Pten^{+/-} mice.

Hollander MC, Balogh AR, Liwanag J, Han W, Linnoila RI, Anver MR, Dennis PA.
Neoplasia 10, 866-872 (2008).

Single nucleotide polymorphisms, apoptosis, and the development of severe late adverse effects after radiotherapy.

Azria D, Ozsahin M, Kramar A, Peters S, Atencio DP, Crompton NE, Mornex F, Pèlegri A, Dubois JB, Mirimanoff RO, Rosenstein BS.
Clin. Cancer Res. 14, 6284-6288 (2008).

Improved identification of von Hippel-Lindau gene alterations in clear cell renal tumors.

Nickerson ML, Jaeger E, Shi Y, Durocher JA, Mahurkar S, Zaridze D, Matveev V, Janout V, Kollarova H, Bencko V, Navratilova M, Szeszenia-Dabrowska N, Mates D, Mukeria A, Holcatova I, Schmidt LS, Toro JR, Karami S, Hung R, Gerard GF, Linehan WM, Merino M, Zbar B, Boffetta P, Brennan P, Rothman N, Chow WH, Waldman FM, Moore LE.
Clin Cancer Res. 14, 4726-34 (2008).

Mutations in the LKB1 tumour suppressor are frequently detected in tumours from Caucasian but not Asian lung cancer patients.

Koivunen JP, Kim J, Lee J, Rogers AM, Park JO, Zhao X, Naoki K, Okamoto I, Nakagawa K, Yeap BY, Meyerson M, Wong KK, Richards WG, Sugarbaker DJ, Johnson BE, Jänne PA.
Br. J. Cancer. 99, 245-52 (2008).

TP53 mutations and survival in squamous-cell carcinoma of the head and neck.

Poeta ML, Manola J, Goldwasser MA, Forastiere A, Benoit N, Califano JA, Ridge JA, Goodwin J, Kenady D, Saunders J, Westra W, Sidransky D, Koch WM.
N. Engl. J. Med. 357, 2552-61 (2007).

Surveyor nuclease-based detection of p53 gene mutations in haematological malignancy.

Mitani N, Niwa Y, Okamoto Y.
Ann. Clin. Biochem. 44, 557-9 (2007).

KIT-negative undifferentiated endometrial sarcoma with the amplified epidermal growth factor receptor gene showing a temporary response to Imatinib mesylate.

Mitsuhashi T, Nakayama M, Sakurai S, Fujimura M, Shimizu Y, Ban S, Ogawa F, Hirose T, Ishihara O, Shimizu M.
Ann. Diagn. Pathol. 11, 49-54 (2007).

Allelic dilution obscures detection of a biologically significant resistance mutation in EGFR -amplified lung cancer.

Engelman JA, Mukohara T, Zejnullahu K, Lifshits E, Borrás AM, Gale CM, Naumov GN, Yeap BY, Jarrell E, Sun J, Tracy S, Zhao X, Heymach JV, Johnson BE, Cantley LC, Jänne PA.
J. Clin. Invest. 116, 2695-2706 (2006).

Erlotinib for frontline treatment of advanced non-small cell lung cancer: a phase II study.

Giaccone G, Gallegos Ruiz M, Le Chevalier T, Thatcher N, Smit E, Rodríguez J, Jänne P, Oulid-Aissa D, Soria J.
Clin. Cancer Res. 12, 6049-6055 (2006).

Genetic predictors of adverse radiotherapy effects: the Gene-PARE project.

Ho A, Atencio D, Peters S, Stock R, Formenti S, Cesaretti J, Green S, Haffty B, Drumea K, Leitzin L, Kuten A, Azria D, Ozsahin M, Overgaard J, Andreassen C, Trop C, Park J, Rosenstein B.
Int. J. Radiat. Oncol. Biol. Phys. 65, 646-655 (2006).

Response and resistance in a non-small-cell lung cancer patient with an epidermal growth factor receptor mutation and leptomeningeal metastases treated with high-dose gefitinib.

Jackman D, Holmes A, Lindeman N, Wen P, Kesari S, Borrás A, Bailey C, de Jong F, Jänne P, Johnson B.
J. Clin. Oncol. 24, 4517-4520 (2006).

Exon 19 deletion mutations of epidermal growth factor receptor are associated with prolonged survival in non-small cell lung cancer patients treated with Gefitinib or Erlotinib.

Jackman D, Yeap B, Sequist L, Lindeman N, Holmes A, Joshi V, Bell D, Huberman M, Halmos B, Rabin M, Haber D, Lynch T, Meyerson M, Johnson B, Jänne P.
Clin. Cancer Res. 12, 3908-3914 (2006).

The JAK2 V617F mutation occurs in hematopoietic stem cells in polycythemia vera and predisposes toward erythroid differentiation.

Jamieson C, Gotlib J, Durocher J, Chao M, Mariappan M, Lay M, Jones C, Zehnder J, Lilleberg S, Weissman I.
Proc. Natl. Acad. Sci. 103, 6224-6229 (2006).

A rapid and sensitive enzymatic method for epidermal growth factor receptor mutation screening.

Jänne P, Borrás A, Kuang Y, Rogers A, Joshi V, Liyanage H, Lindeman N, Lee J, Halmos B, Maher E, Distel R, Meyerson M, Johnson B.
Clin. Cancer Res. 12, 751-758 (2006).

Effect of epidermal growth factor receptor tyrosine kinase domain mutations on the outcome of patients with non-small cell lung cancer treated with epidermal growth factor receptor tyrosine kinase inhibitors.

Jänne P, Johnson B.
Clin. Cancer Res. 12, 4416s-4420s (2006).

Oncology continued

Immunohistochemical evaluation of KIT expression in sarcomas of the gynecologic region.

Nakayama M, Mitsushashi T, Shimizu Y, Ban S, Ogawa F, Ishihara O, Shimizu M.
Int. J. Gynecol. Pathol. 25, 70-76 (2006).

Mutation analysis of hCDC4 in AML cells identifies a new intronic polymorphism.

Nowak D, Mossner M, Baldus C, Hopfer O, Hofmann W.
Int. J. Med. Sci. 3, 148-151 (2006).

A sensitive high-throughput method to detect activating mutations of Jak2 in peripheral-blood samples.

Sattler M, Walz C, Crowley B, Lengfelder E, Jänne P, Rogers A, Kuang Y, Distel R, Reiter A, Griffin J.
Blood 107, 1237-1238 (2006).

Activity of the tyrosine kinase inhibitor PKC412 in a patient with mast cell leukemia with the D816V KIT mutation.

Gotlib J, Berube C, Growney J, Chen C, George T, Williams C, Kajiguchi T, Ruan J, Lilleberg S, Durocher J, Lichy J, Wang Y, Cohen P, Arber D, Heinrich M, Neckers L, Galli S, Gilliland D, Coutre S.
Blood 106, 2865-2870 (2005).

Germline Genetic Screening

A molecular analysis of mutations at the complex dumpy locus in *Drosophila melanogaster*.

Carmon A, Guertin M, Grushko O, Marshall B, MacIntyre R.
PLoS ONE 5, e12319 (2010).

Meganucleases can restore the reading frame of a mutated dystrophin.

Chapdelaine P, Pichavant C, Rousseau J, Pâques F, Tremblay J.
Gene Ther. 17, 846-58 (2010).

Screening for mutations in kidney-related genes using SURVEYOR nuclease for cleavage at heteroduplex mismatches.

Voskarides K, Deltas C.
J. Mol. Diagn. 11, 311-318 (2009).

Rapid screening for nuclear genes mutations in isolated respiratory chain complex I defects.

Pagniez-Mammeri H, Lombes A, Brivet M, Ogier-de Baulny H, Landrieu P, Legrand A, Slama A.
Mol. Genet. Metab. 96, 196-200 (2009).

Novel method for genomic analysis of PKD1 and PKD2 mutations in autosomal dominant polycystic kidney disease.

Tan YC, Blumenfeld JD, Anghel R, Donahue S, Belenkaya R, Balina M, Parker T, Levine D, Leonard DG, Rennert H.
Hum. Mutat. 30 264-273 (2009).

Comparison of the mismatch-specific endonuclease method and denaturing high-performance liquid chromatography for the identification of HBB gene mutations.

Hung C, Su Y, Lin C, Chang Y, Chang C, Cheng W, Chen C, Lee C, Lin W.
BMC Biotechnol. 8, 62-70 (2008).

NPHS2 screening with SURVEYOR in Hellenic children with steroid-resistant nephrotic syndrome.

Voskarides K, Makariou C, Papagregoriou G, Stergiou N, Printza N, Alexopoulos E, Elia A, Papachristou F, Pierides A, Georgaki E, Deltas C.
Pediatr. Nephrol. 23, 1373-1375 (2008).

Genetic regulation of beta-ureidopropionase and its possible implication in altered uracil catabolism.

Thomas H, Ezzeldin H, Guarcello V, Mattison L, Fridley B and Diasio R.
Pharmacogenet. Genomics 18; 25-35 (2008).

Screening human genes for small alterations performing an enzymatic cleavage mismatched analysis (ECMA) protocol.

Vogiatzakis N, Kekou K, Sophocleous C, Kitsiou S, Mavrou A, Bakoula C, Kanavakis E.
Mol. Biotechnol. 37 12-9 (2007).

Genetic regulation of dihydropyrimidinase and its possible implication in altered uracil catabolism.

Thomas H, Ezzeldin H, Guarcello V, Mattison L, Fridley B, Diasio R.
*Pharmacogenet Genomics. Nov;*17(11):973-87 (2007).

Development of a rapid, reliable genetic test for Pseudoxanthoma Elasticum.

Shi Y, Terry S, Terry P, Bercovitch L, Gerard G.
J. Mol. Diagn. 9, 105-112 (2007).

A new detection method for ATRX gene mutations using a mismatch-specific endonuclease.

Wada T, Fukushima Y, Saitoh S.
Am. J. Med. Genet. A 140, 1519-1523 (2006).

Mitochondrial DNA Analysis

The link between mitochondrial DNA hypervariable segment I heteroplasmy and ageing among genetically unrelated Latvians.

Pliss L, Brakmanis A, Ranka R, Elferts D, Krumina A, Baumanis V.
Exp Gerontol. 46, 560-8 (2011).

Mitochondrial DNA damage and repair in RPE associated with aging and age-related macular degeneration.

Lin H, Xu H, Liang F, Liang H, Gupta P, Havey A, Boulton M, Godley B.
Invest. Ophthalmol. Vis. Sci. 52 3521-3529 (2011).

Surveyor nuclease detection of mutations and polymorphisms of mtDNA in children.

Pilch J, Asman M, Jamroz E, Kajor M, Kotrys-Puchalska E, Goss M, Krzak M, Witecka J, Gmiński J, Sieroń AL.
Pediatr. Neurol. 43, 325-330 (2010).

Strategy in diagnosis of mitochondrial diseases.

Lebre A.
Pathol. Biol (Paris) 58, 353-356 (2009).

Parkinson's disease brain mitochondria have impaired respirasome assembly, age-related increases in distribution of oxidative damage to mtDNA and no differences in heteroplasmic mtDNA mutation abundance.

Arthur C, Morton S, Dunham L, Keeney P, Bennett J Jr.
Mol. Med. Reports 4, 37-49 (2009).

Rapid identification of mitochondrial DNA (mtDNA) mutations in neuromuscular disorders by using surveyor strategy.

Bannwarth S, Procaccio V, Rouzier C, Fragaki K, Poole J, Chabrol B, Desnuelle C, Pouget J, Azulay J, Attarian S, Pellissier J, Gargus J, Abdenur J, Mozaffar T, Calvas P, Labauge P, Pages M, Wallace D, Lambert J and Paquis-Flucklinger V.
Mitochondrion 8, 136-145 (2008).

Mitochondrial DNA Analysis continued

Rapid identification of unknown heteroplasmic mutations across the entire human mitochondrial genome with mismatch-specific Surveyor nuclease.

Bannwarth S, Procaccio V, Paquis-Flucklinger V. *Nat. Protoc.* 1, 2037-2047 (2006).

Surveyor™ Nuclease: A new strategy for a rapid identification of heteroplasmic mitochondrial DNA mutations in patients with respiratory chain defects.

Bannwarth S, Procaccio V, Paquis-Flucklinger V. *Hum. Mutat.* 25, 575-582 (2005).

Plant Biology

Site-directed mutagenesis in *Arabidopsis* using custom-designed zinc finger nucleases.

Osakabe K, Osakabe Y, Toki S. *Proc. Natl. Acad. Sci.* 107, 12034-12039 (2010).

A Modified TILLING Method for Wheat Breeding.

Dong C, Dalton-Morgan J, Vincent K, Sharp P. *The Plant Genome* 2, 39-47 (2009).

TILLING in the two-rowed barley cultivar 'Barke' reveals preferred sites of functional diversity in the gene HvHox1.

Gottwald S, Bauer P, Komatsuda T, Lundqvist U, Stein N. *BMC Mol. Biol.* 2, 258-271 (2009).

CEL I Nuclease Digestion for SNP Discovery and Marker Development in Common Bean (*Phaseolus vulgaris* L.).

Galeano C, Gomez M, Rodriguez L, Blair M. *Croat. Med. J.* 49, 381-394 (2009).

Self-EcoTILLING to identify single-nucleotide mutations in multigene family.

Wang G-X, Imaizumi T, Li W, Saitoh H, Terauchi R, Ohsako T, Tominaga T. *Pest Biochem. Physiol.* 92, 24-29 (2008).

TILLMore, a resource for the discovery of chemically induced mutants in barley

Talamè V, Bovina R, Sanguineti M, Tuberosa R, Lundqvist U, Salvi S. *Plant Biotechnol. J.* 6, 477-485 (2008).

A structured mutant population for forward and reverse genetics in Barley (*Hordeum vulgare* L.).

Caldwell D, McCallum N, Shaw P, Muehlbauer G, Marshall D, Waugh R. *Plant J.* 40, 143-150 (2004).

Heteroduplex detection with a plant DNA endonuclease for standard gel electrophoresis.

Scafino M, Pilotto A, Papadimitriou S, Sbalzarini M, Ansaldi S, Diegoli M, Porcu E, Grasso M, Brega A, Arbustini E. *Transgenics* 4, 157-166 (2004).

Microbiology

Generation of targeted *Chlamydia trachomatis* null mutants

Kari L, Goheen M, Randall L, Taylor L, Carlson J, Whitmire W, Virok D, Rajaram K, Endresz V, McClarty G, Nelson D, Caldwell H. *Proc Natl Acad Sci U S A.* 108 7189-93 (2011).

Temperature-mediated heteroduplex analysis for the detection of drug-resistant gene mutations in clinical isolates of *Mycobacterium tuberculosis* by denaturing HPLC, SURVEYOR nuclease.

Shi R, Otomo K, Yamada H, Tatsumi T, Sugawara I. *Microbes Infect.* 8, 128-135 (2006).

Localization, mobility and fidelity of retrotransposed Group II introns in rRNA genes.

Conlan L, Stanger M, Ichiyonagi K, Belfort M. *Nucleic Acids Res.* 33, 5262-5270 (2005).

SURVEYOR Nuclease use with TALE Nucleases

A transcription activator-like effector toolbox for genome engineering

Sanjana N; Cong L; Zhou Y, Cunniff M, Feng G, Zhang F *Nat Protoc* 7 171-192 (2012).

Knockout rats generated by embryo microinjection of TALENs

Tesson L, Usal C, Ménoret S, Leung E, Niles B, Remy S, Santiago Y, Vincent A, Meng X, Zhang L, Gregory P, Anegon I, Cost G *Nat Biotechnol.* 29, 695–696 (2011).

A TALE nuclease architecture for efficient genome editing

Miller J, Tan S, Qiao G, Barlow KA, Wang J, Xia D, Meng X, Paschon D, Leung E, Hinkley S, Dulay G, Hua K, Ankoudinova I, Cost G, Urnov F, Zhang H, Holmes M, Zhang L, Gregory P, Rebar E. *Nat. Biotechnol.* 29, 143-148 (2011).

SURVEYOR Nuclease use with Zinc Finger Nucleases

Engineering HIV-Resistant Human CD4+ T Cells with CXCR4-Specific Zinc-Finger Nucleases

Wilén CB, Wang J, Tilton JC, Miller JC, Kim KA, et al. *PLoS Pathog* 7, e1002020 (2011)

DNA Ligase III Promotes Alternative Nonhomologous End-Joining during Chromosomal Translocation Formation.

Simsek D, Brunet E, Wong SY-W, Katyal S, Gao Y, et al. *PLoS Genet* 7, e1002080. (2011)

Efficient generation of a biallelic knockout in pigs using zinc-finger nucleases

Hauschild J, Petersen B, Santiago Y, Queisser A, Carnwath J, Lucas-Hahn A, Zhang L, Meng X, Gregory P, Schwinzer R, Cost G, Niemann H. *Proc Natl Acad Sci U S A.* 108, 12013-7 (2011).

Gene Editing of Human Embryonic Stem Cells via an Engineered Baculoviral Vector Carrying Zinc-finger Nucleases

Lei Y, Lee C-L, Joo K-I, Zarzar J, Liu Y, Dai B, Fox V, Wang P. *Mol Ther.* 19, 942-950 (2011).

Gene targeting to the ROSA26 locus directed by engineered zinc finger nucleases.

Perez-Pinera P, Ousterout D, Brown M, Gersbach C. *Nucleic Acids Res.* 2011 Dec 14. [Epub ahead of print]

Transient cold shock enhances zinc-finger nuclease-mediated gene disruption

Doyon Y, Choi V, Xia D, Vo T, Gregory P, Holmes M. *Nat Methods* 7, 459-460 (2010).

Knockout of exogenous EGFP gene in porcine somatic cells using zinc-finger nucleases

Watanabe M, Umeyama K, Matsunari H, Takayanagi S, Haruyama E, Nakano K, Fujiwara T, Ikezawa Y, Nakauchi H, Nagashima H. *Biochem. Biophys. Res. Commun.* 402, 14-18 (2010).

SURVEYOR Nuclease use with Zinc Finger Nucleases continued

Generation of gene-specific mutated rats using zinc-finger nucleases

Geurts A, Cost G, Rémy S, Cui X, Tesson L, Usal C, Ménoret S, Jacob H, Anegon I, Buelow R.
Methods Mol. Biol. 597, 211-225 (2010).

Knockout rats via embryo microinjection of zinc-finger nucleases

Geurts A, Cost G, Freyvert Y, Zeitler B, Miller J, Choi V, Jenkins S, Wood A, Cui X, Meng X, Vincent A, Lam S, Michalkiewicz M, Schilling R, Foeckler J, Kalloway S, Weiler H, Ménoret S, Anegon I, Davis G, Zhang L, Rebar E, Gregory P, Urnov F, Jacob J, Buelow R.
Science 325, 433 (2009).

Synthetic Gene Error Correction

Error correction of microchip synthesized genes using Surveyor nuclease

Saaem I, Ma S, Quan J, Tian J.
Nucleic Acids Res. Nov 29 (2011). [Epub ahead of print]

Parallel on-chip gene synthesis and application to optimization of protein expression

Quan J, Saaem I, Tang N, Ma S, Negre N, Gong H, White KP, Tian J.
Nat Biotechnol. 29 449-52 (2011).

Endonucleases: tools to correct the dystrophin gene

Rousseau J, Chapdelaine P, Boisvert S, Almeida L, Corbeil J, Montpetit A, Tremblay J.
J Gene Med. 13, 522-37 (2011).

SURVEYOR Nuclease Technology

A highly sensitive and specific biosensor for ligation- and PCR-free detection of MicroRNAs

Gao Z, Peng Y.
Biosens Bioelectron. 26, 3768-3773 (2011).

s-RT-MELT: a novel technology for mutation screening

Li J, Makrigiorgos GM.
Methods Mol Biol. 653, 207-19 (2010).

A rapid and general assay for monitoring endogenous gene modification

Guschin D, Waite A, Katibah G, Miller J, Holmes M, Rebar E.
Methods Mol Biol. 649, 247-56 (2010).

High-throughput identification of mutations using a combination of CEL I fragmentation and SAGE technology

Zhang C, Li Y, Wang X, Zhang L, Li X, Wang Y, Xu S.
Genet Test Mol Biomarkers 13, 97-103 (2009).

Endonucleolytic mutation analysis by internal labeling (EMAIL)

Cross M, Waters D, Lee L, Henry R.
Electrophoresis 29, 1291-1301 (2008).

Development of a simple and highly sensitive mutation screening system by enzyme mismatch cleavage with optimized conditions for standard laboratories

Tsuji T, Niida Y.
Electrophoresis 29, 1473-83 (2008).

s-RT-MELT for rapid mutation scanning using enzymatic selection and real time DNA-melting: new potential for multiplex genetic analysis

Li J, Berbeco R, Distel R, Jänne P, Wang L, Makrigiorgos GM.
Nucleic Acids Res. 35, e84 (2007).

Surveyor nuclease-based genotyping of SNPs.

Mitani N, Tanaka S, Okamoto Y.
Clin. Lab. 52, 385-6 (2006).

A method for clone sequence confirmation using a mismatch-specific DNA endonuclease.

Qiu P, Shandilya H, Gerard GF.
Mol. Biotechnol. 29, 11-18 (2005).

Improving specificity of DNA hybridization-based methods.

Chalaya T, Gogvadze E, Buzdin A, Kovalskaya E, Sverdlov ED.
Nucleic Acids Res. 32, e130 (2004).

Mutation detection using Surveyor™ nuclease.

Qiu P, Shandilya H, D'Alessio JM, O'Connor K, Durocher J, Gerard GF.
Biotechniques 36, 702-707 (2004).



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