ACCELERATING PROGRESS IS IN OUR GENES

PUBLICATIONS LIST

HALOPLEX TARGET ENRICHMENT SYSTEM

Cancer Research

1. Aihara K, Mukasa A, Gotoh K, Saito K, Nagae G, Tsuji S, Tatsuno K, Yamamoto S, Takayanagi S, Narita Y, Shibui S, Aburatani H and Saito N. **H3F3A K27M mutations in thalamic gliomas from young adult patients**. *Neuro Oncol*. (2014) 16(1): 140-6
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2. Mansouri L, Sutton LA, Ljungström V, Sörqvist EF, Gunnarsson R, Smedby KE, Juliusson G, Stamatopoulos K, Nilsson M and Rosenquist R. **Feasibility of targeted next-generation sequencing of the TP53 and ATM genes in chronic lymphocytic leukemia**. *Leukemia*. (2014) 28(3):694-6
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3. Menezes J, Makishima H, Gomez I, Acquadro F, Gómez-López G, Graña O, Dopazo A, Alvarez S, Trujillo M, Pisano DG, Maciejewski JP and Cigudosa JC. **CSF3R T618I co-occurs with mutations of splicing and epigenetic genes and with a new PIM3 truncated fusion gene in chronic neutrophilic leukemia**. *Blood Cancer J*. (2013) 3: e158
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4. Berglund EC, Lindqvist CM, Hayat S, Övernäs E, Henriksson N, Nordlund J, Wahlberg P, Forestier E, Lönnerholm G and Syvänen AC. **Accurate detection of subclonal single nucleotide variants in whole genome amplified and pooled cancer samples using HaloPlex target enrichment**. *BMC Genomics*. (2013) 14:856
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5. Schulz E, Velntin A, Ulz P, Beham-Schmid C, Lind K, Rupp V, Lackner H, Wolfler A, Zebisch A, Olipitz W, Geigl J, Berghold A, Speicher MR and Sill H. **Germline Mutations in the DNA damage Response Genes BRCA1, BRCA2, BARD1 and TP53 in Patients with Therapy Related Myeloid Neoplasms**. *J Med. Genet*. (2012) 49: 422-428
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6. Oike T, Ogiwara H, Tominaga Y, Ito K, Ando O, Tsuta K, Mizukami T, Shimada Y, Isomura H, Komachi M, Furuta K, Watanabe SI, Nakano T, Yokota J and Kohno T. **A Synthetic Lethality-Based Strategy to Treat Cancers Harboring a Genetic Deficiency in the Chromatin Remodling Factor BRG1**. *Cancer Res*. (2013) 73(17): 5508-18
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7. Balbas-Martinez C, Sagrera A, Carillo-de-Santa-Pau E, Earl J, Marquez M, Vazquez M, Lapi E, Castro-Giner F, Beltran S, Bayes M, Carrato A, Cigudosa JC, Dominguez O, Gut M, Herranz J, Juanpere N, Kogevinas M, LAnga X, Lopez-Knowles E, Lorente JA, Lloreta J, Pisano DG, Richart L, Rico D, Salgado RN, Tardon A, Chanock S, Heath S, Valencia A, Losada A, Gut I, Malats N and Real FX. **Recurrent Inactivation of STAG2 in Bladder Cancer is not Associated with Aneuploidy**. *Nature Genetics* (2013)
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8. Mitchell G, Ballinger ML, Wong S, Hewitt C, James P, Young M, Cipponi A, Pang T, Goode DL, Dobrovic A, Thomas DM. **High Frequency of Germline TP53 Mutations in a Prospective Adult-Onset Sarcoma Cohort**. *PLOS One* (2013) 8(7): e69026
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9. Koopmans AE, Vaarwater J, Paridaens D, Naus NC, Kilic E, Klein A. **Patient Survival in Uveal Melanoma is not affected by Oncogenic Mutations in GNAQ and GNA11**. *British J. Cancer* (2013) 109: 493-496
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Inherited Disease Research

1. Ylikallio E, Johari M, Konovalova S, Moilanen JS, Kiuru-Enari S, Auranen M, Pajunen L and Tyynismaa H. **Targeted Next-Generation Sequencing Reveals Further Genetic Heterogeneity in Axonal Charcot-Marie-Tooth Neuropathy and A Mutation in HSPB1**. *Eur. J. Hum. Genet*. (2013) 1-6
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2. Tsurusaki Y, Okamoto N, Ohashi H, Mizuno S, Matsumoto N, Makita Y, Fukuda M, Isidor B, Perrier J, Aggarwal S, Dalal AB, Al0Kindy A, Liebelt J, Mowat D, Nakashima M, Saitsu H, Miyake N and Masumoto N. **Coffin-Siris Syndrome is a SWI/SNF Complex Disorder**. *Clin. Genet*. (2013)
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3. Braun TA, Mullins RF, Wagner AH, Andorf JL, Johnston RM, Bakall BB, Deluca AP, Fishman GA, Lam, BL, Weleber RG, Cideciyan AV, Jacobson AG, Sheffield VC, Tucker BA and Stone EM. **Non-Exomic and Synonymous Variants in ABCA4 are and Important Cause of Stargardt Disease**. *Hum. Molec. Genet*. (2013)
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Other

1. Suzuki E, Yatsuga S, Igarashi M, Miyado M, Nakabayashi K, Hayashi K, Hata K, Umezawa A, Yamada G, Ogata T and Fukami M. **De novo Frameshift Mutation in Fibroblast Growth Factor 8 in a Male Patient with Gonadotropin Deficiency**. *Horm Res Paediatr*. (2014) 81(2):139-44
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