**4467** Publications using Droplet Digital PCR

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	1 Dec, 2014

12p microRNA expression in fibroblast cell lines from probands with Pallister-Killian syndrome

E Chromosome Res.

Q Izumi, K;Zhang, Z;Kaur, M;Krantz, ID;
Pallister-Killian syndrome is a multisystem sporadic genetic diagnosis characterized by facial dysmorphia, variable developmental delay and intellectual impairment, hypotonia, seizures, diaphragmatic hernia, and other systemic abnormalities. Pallister-Killian syndrome is typically caused by the presence of a supernumerary isochromosome that is always present in a tissue limit... Imore

**16S rDNA droplet digital**13 Nov, 2019

PCR for monitoring

bacterial DNAemia in

bloodstream infections

PLoS ONE

② Ziegler, I;Lindström, S;Källgren, M;StrÃ¥lin, K;Mölling, P;

Repeated quantitative measurement of bacterial DNA on whole blood has been shown to be a promising method for monitoring bloodstream infection (BSI) with selected bacterial species. To enable broad use of this method, we developed a quantitative droplet digital PCR (ddPCR) method for 16S rDNA. It was validated with species-specific ddPCRs for Staphylococcus au... 1 more

Reproduction, Fertility and Development

♠ Rho, NY;Favetta, LA;Mogas, T;King, WA; Testis-specific protein Y (TSPY) is one of the genes located in the male-specific region of the Y chromosome involved in male reproduction, particularly in spermatogenesis. It has been shown to have different copy number (CN) in different species, in different individuals within a species and in the same family (i.e. among brothers from the same father). Previous studies ... \textstyle{\textstyle{1}}\text{more}

1q21.1 microduplication: 22 Aug, 2018 large verbal-nonverbal performance discrepancy and ddPCR assays of HYDIN/HYDIN2 copy number

- NPJ Genom Med
- ② Xavier, J;Zhou, B;Bilan, F;Zhang, X;Gilbert-Dussardier, B;Viaux-Savelon, S;Pattni, R;Ho, SS;Cohen, D;Levinson, DF;Urban, AE;Laurent-Levinson, C;

#### **22q11 deletion syndrome:** 9 Jun, 2015 current perspective.

- Appl Clin Genet
- HacıhamdioÄlu, B;HacıhamdioÄlu, D;Delil, K;

Chromosome 22q11 is characterized by the presence of chromosome-specific low-copy repeats or segmental duplications. This region of the chromosome is very unstable and susceptible to mutations. The misalignment of low-copy repeats during nonallelic homologous recombination leads to the deletion of the 22q11.2 region, which results in 22q11 deletion syndrome (22q11... \textstyle{\textstyle{\textstyle{1}}} \textstyle{\textstyle{1}} \textstyle{\textstyle{1}}

**5-Formylcytosine Could** 6 Dec, 2016 Be a Semipermanent Base in Specific Genome Sites

Angew. Chem. Int. Ed. Engl.

Su, M;Kirchner, A;Stazzoni, S;Mù/₄ller,
M;Wagner, M;Schröder, A;Carell, T;
5-Formyl-2'-deoxycytosine (fdC) is a recently
discovered epigenetic base in the genome of stem
cells, with yet unknown
functions. Sequencing data show that the base is
enriched in CpG islands of promoters and hence
likely involved in the
regulation of transcription during cellular
differentiation. fdC is known to be recognized and
excised by the enzyme thymi... ↓more

6-Phosphofructokinase
1 May, 2017
and ribulose-5-phosphate
3-epimerase in
methylotrophic Bacillus
methanolicus ribulose
monophosphate cycle

🗏 Appl. Microbiol. Biotechnol.

• Le, SB; Heggeset, TMB; Haugen, T; Nærdal, I; Brautaset, T;

D-Ribulose-5-phosphate-3-epimerase (RPE) and 6-phosphofructokinase (PFK) catalyse two reactions in the ribulose monophosphate (RuMP) cycle in Bacillus methanolicus. The B. methanolicus wild-type strain MGA3 possesses two putative rpe and pfk genes encoded on plasmid pBM19 (rpe1-MGA3 and pfk1-MGA3) and on the chromosome (rpe2-MGA3 and pfk2... \ \textbf{more}

A BAG3 chaperone 20 Jul, 2017 complex maintains cardiomyocyte function during proteotoxic stress

**■** JCI Insight

● Judge, LM;Perez-Bermejo, JA;Truong, A;Ribeiro, AJ;Yoo, JC;Jensen, CL;Mandegar, MA;Huebsch, N;Kaake, RM;So, PL;Srivastava, D;Pruitt, BL;Krogan, NJ;Conklin, BR;

Molecular chaperones regulate quality control in the human proteome, pathways that have been implicated in many diseases,

including heart failure. Mutations in the BAG3 gene, which encodes a co-chaperone protein, have been associated with heart

failure due to both inherited and sporadic dilated cardiomyopathy. Familial BAG3 mutations are autosomal dominant and... 

Imore

A blood-based gene 1 Jan, 2017 expression and signaling pathway analysis to differentiate between high and low grade gliomas

Oncol. Rep.

potential blood-based

• Ponnampalam, SN; Kamaluddin, NR; Zakaria, Z; Matheneswaran, V; Ganesan, D; Haspani, MS; Ryten, M; Hardy, JA;

The aims of the present study were to undertake gene expression profiling of the blood of glioma patients to determine key genetic components of signaling pathways and to develop a panel of genes that could be used as a

biomarker to differentiate between high and low grade gliomas, non-gliomas and control samples. In this study, blood sa... **\\_lore** 

A Blood-based Test for the 5 Apr., 2018
Detection of ROS1 and RET
Fusion Transcripts from

#### Circulating Ribonucleic Acid Using Digital Polymerase Chain Reaction

J Vis Exp

Mellert, HS;Alexander, KE;Jackson, LP;Pestano, GA:

We have developed novel methods for the isolation and characterization of tumor-derived circulating ribonucleic acid (cRNA) for blood-based liquid biopsy. Robust detection of cRNA recovered from blood represents a solution to a critical unmet need in clinical diagnostics. The test begins with the collection of whole blood into blood collection tubes containing preservatives... 1more

## A Burden of Rare Variants 4 Feb, 2016 Associated with Extremes of Gene Expression in Human Peripheral Blood.

Am. J. Hum. Genet.

② Zhao, J;Akinsanmi, I;Arafat, D;Cradick, TJ;Lee, CM;Banskota, S;Marigorta, UM;Bao, G;Gibson, G; In order to evaluate whether rare regulatory variants in the vicinity of promoters are likely to impact gene expression, we

conducted a novel burden test for enrichment of rare variants at the extremes of expression. After sequencing 2-kb promoter regions of 472 genes in 410 healthy adults, we performed a quadratic regression of rare variant count on bins of peripher... 1more

## A C9ORF72 BAC mouse 12 Jun, 2017 model recapitulates key epigenetic perturbations of ALS/FTD

■ Mol Neurodegener

Sesanov, R; Cabrera, GT; Andrade, NS; Gendron, TF; Brown, RH; Benatar, M; Wahlestedt, C; Mueller, C; Zeier, Z;

Amyotrophic Lateral Sclerosis (ALS) is a fatal and progressive neurodegenerative disorder with identified genetic causes representing a significant minority of all cases. A GGGCC hexanucleotide repeat expansion (HRE) mutation within the C9ORF72 gene has recently been identified as the most frequent known cause of ALS. The expansion leads to partial h... 1more

# A cAMP/PKA/Kinesin-1 23 Apr, 2018 Axis Promotes the Axonal Transport of Mitochondria in Aging Drosophila Neurons

- Curr. Biol.
- Vagnoni, A;Bullock, SL;

Mitochondria play fundamental roles within cells, including energy provision, calcium homeostasis, and the regulation of apoptosis. The transport of mitochondria by microtubule-based motors is critical for neuronal structure and function. This process allows local requirements for mitochondrial functions to be met and also facilitates recycling of these organelles [1, 2]. ... \textstyle more

#### A Case Study of Genomic 6 Nov, 2018 Instability in an Industrial Strain of Saccharomyces cerevisiae

- **G**3 (Bethesda)
- Rodrigues-Prause, A;Sampaio, NMV;Gurol, TM;Aguirre, GM;Sedam, HNC;Chapman, MJ;Malc, EP;Ajith, VP;Chakraborty, P;Tizei, PA;Pereira, GAG;Mieczkowski, PA;Nishant, KT;Argueso, JL; The Saccharomyces cerevisiae strain JAY270/PE2 is a highly efficient biocatalyst used in the production of bioethanol from sugarcane feedstock. This strain is heterothallic and diploid, and its genome is characterized by abundant structural and nucleotide polymorphisms between homologous

chromosomes. One of the reasons it is favored by many distilleries is th... **\\_more** 

A CD19/CD3 bispecific 9 May, 2018 antibody for effective immunotherapy of chronic lymphocytic leukemia in the ibrutinib era

Blood

Robinson, HR;Qi, J;Cook, EM;Nichols, C;Dadashian, EL;Underbayev, C;Herman, SEM;Saba, NS;Keyvanfar, K;Sun, C;Ahn, IE;Baskar, S;Rader, C;Wiestner, A;

The Bruton's tyrosine kinase inhibitor ibrutinib induces high rates of clinical response in chronic lymphocytic leukemia (CLL).

However, there remains a need for adjunct treatments to deepen response and to overcome drug resistance. Blinatumomab, a CD19/CD3 bispecific antibody (bsAb) designed in the BiTE® format, is FDA approved for the treatment of relapsed or r... \textstyle \textstyle

A Cell-free DNA Barcode5 Jun, 2019
Enabled Single-Molecule Test
for Noninvasive Prenatal
Diagnosis of Monogenic
Disorders: Application to βThalassemia

Adv Sci (Weinh)

● Yang, X;Zhou, Q;Zhou, W;Zhong, M;Guo, X;Wang, X;Fan, X;Yan, S;Li, L;Lai, Y;Wang, Y;Huang, J;Ye, Y;Zeng, H;Chuan, J;Du, Y;Ma, C;Li, P;Song, Z;Xu, X;

Noninvasive prenatal testing of common aneuploidies has become routine over the past decade, but testing of monogenic disorders remains a challenge in clinical implementation. Most recent studies have inherent limitations, such as complicated procedures, a lack of versatility, and the need for prior knowledge of parental genotypes or haplotypes. To overcome the... 1more

Cellâfree DNA 1 Apr, 2019 **BarcodeâEnabled** SingleâMolecule Test for **Noninvasive Prenatal** Diagnosis of Monogenic **Disorders: Application** βâThalassemia

- Adv. Sci.
- Yang, X;Zhou, Q;Zhou, W;Zhong, M;Guo, X;Wang, X;Fan, X;Yan, S;Li, L;Lai, Y;Wang, Y;Huang, J;Ye, Y;Zeng, H;Chuan, J;Du, Y;Ma, C;Li, P;Song, Z;Xu, X;

Noninvasive prenatal testing of common aneuploidies has become routine over the past decade, but testing of monogenic disorders remains a challenge in clinical implementation. Most recent studies have inherent limitations, such as complicated procedures, a lack of versatility, and the need for prior knowledge of parental genotypes or haplotypes. To overcome the... 1more

A cetuximab-mediated 12 Mar, 2019 suicide system in CAR-modified hematopoietic stem cells for cancer therapy

- Hum. Gene Ther.
- Skao, RL;Truscott, LC;Chiou, TT;Tsai, W;Wu, AM;De Oliveira, SN;

We propose using gene-modification of HSC to create persistent generation of multilineage immune effectors to directly target cancer cells. Gene-modified human hematopoietic

stem cells (HSC) have been used to introduce

genes to correct, prevent or treat diseases. Concerns regarding malignant transformation, abnormal hematopoiesis and autoimmunity exist, making t... 1more

A Circulating microRNA 2 Jan, 2017
Signature Predicts AgeBased Development of
Lymphoma

PLoS ONE

Beheshti, A; Vanderburg, C; McDonald, JT; Ramkumar, C; Kadungure, T; Zhang, H; Gartenhaus, RB; Evens, AM;

Extensive epidemiological data have demonstrated an exponential rise in the incidence of non-Hodgkin lymphoma (NHL) that is associated with increasing age. The molecular etiology of this remains largely unknown, which impacts the effectiveness of treatment for patients. We proposed that age-dependent circulating microRNA (miRNA) signatures in the host influence ... 1 more

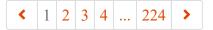
A Clamp-Based One-Step
Droplet Digital Reverse
Transcription PCR (ddRT-PCR) for Precise
Quantitation of Messenger
RNA Mutation in Single
Cells

ACS Sens

Sun, Y;Tian, H;Liu, C;Yang, D;Li, Z;

Precise detection of the low copy numbers of messenger RNA (mRNA) mutation in single cells is of great significance but still remains challenging. Herein, by integrating the outstanding features of a rationally designed peptide nucleic acid (PNA) clamp for highly selective discrimination of single-nucleotide

variation, and droplet digital PCR for ultrasensitive and precise quantif...



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