gene therapy

	NCT Number	Title	Authors	Description	Identifier	Dates
1	pubmed:36118672	Pancancer Analysis of Revealed *TDO2* as a Biomarker of Prognosis and Immunotherapy	Jing Cui Yongjie Tian Tianhang Liu Xueyan Lin Lanyu Li Zhonghui Li Liang Shen	CONCLUSION: Therefore, our results suggest that TDO2 can function as a potential prognostic biomarker due to its role in tumor immunity regulation.	pmid:36118672 pmc:PMC9481368 doi:10.1155/2022/5447017	Mon, 19 Sep 2022 06:00:00 -0400
2	pubmed:36119068	A robust CD8^{+} T cell-related classifier for predicting the prognosis and efficacy of immunotherapy in stage III lung adenocarcinoma	Jinteng Feng Longwen Xu Shirong Zhang Luying Geng Tian Zhang Yang Yu Rui Yuan Yusheng He Zhuhui Nan Min Lin Hui Guo	Patients with stage III lung adenocarcinoma (LUAD) have significant survival heterogeneity, meanwhile, CD8^(+) T cell has a remarkable function in immunotherapy. Therefore, developing novel biomarkers based on CD8^(+) T cell can help evaluate the prognosis and guide the strategy of immunotherapy for patients with stage III LUAD. Thus, we abstracted twelve datasets from multiple online databases and grouped the stage III LUAD patients into training and validation sets. We then used WGCNA and...	pmid:36119068 pmc:PMC9471021 doi:10.3389/fimmu.2022.993187	Mon, 19 Sep 2022 06:00:00 -0400
3	pubmed:36119097	Gain-of-function defects in toll-like receptor 8 shed light on the interface between immune system and bone marrow failure disorders	Jack Bleesing	In this article, we will share lessons that patients with gain-of-function defects in Toll-like receptor 8 (TLR8-GOF) can teach us about the interface between bone marrow failure (BMF) disorders and inborn errors of immunity (IEI), subsequently referred to as "Interface Disorders". TLR8-GOF is a relatively young entity (from a discovery standpoint) that-through both similar and dissimilar disease characteristics-can increase our understanding of interface disorders, for example, as it pertains...	pmid:36119097 pmc:PMC9479092 doi:10.3389/fimmu.2022.935321	Mon, 19 Sep 2022 06:00:00 -0400
4	pubmed:36119110	Environmental factors influencing the risk of ANCA-associated vasculitis	Wen-Man Zhao Zhi-Juan Wang Rui Shi Yu-Yu Zhu Sen Zhang Rui-Feng Wang De-Guang Wang	Antineutrophil cytoplasmic antibody (ANCA)-associated vasculitis (AAV) is a group of diseases characterized by inflammation and destruction of small and medium-sized blood vessels. Clinical disease phenotypes include microscopic polyangiitis (MPA), granulomatosis with polyangiitis (GPA), and eosinophilic granulomatosis with polyangiitis (EGPA). The incidence of AAV has been on the rise in recent years with advances in ANCA testing. The etiology and pathogenesis of AAV are multifactorial and...	pmid:36119110 pmc:PMC9479327 doi:10.3389/fimmu.2022.991256	Mon, 19 Sep 2022 06:00:00 -0400
5	pubmed:36119920	Deep Fusion of Intelligent Meridian Sensing Technology and Huoluo Xiaoling Pills in the Treatment of Knee Osteoarthritis	Jiye Sun Ziou Wang Piao Wu Jinxuan Wei Xiaowei Sun Hongtao Li	Based on the deep fusion of intelligent meridian sensing technology and Huoluo Xiaoling Pill (HXP) in the treatment of knee osteoarthritis (KOA), firstly, the effective components and targets of Salvia miltiorrhiza, Angelica sinensis, frankincense, and myrrh were obtained by using TCMSP, SwissADME, and Swisstarget databases. Similarly, relevant targets of KOA were collected through GeneCards, OMIM, TTD, PharmGKB, and DrugBank databases. Next, the potential targets of ZXP in the treatment of KOA...	pmid:36119920 pmc:PMC9473892 doi:10.1155/2022/8043674	Mon, 19 Sep 2022 06:00:00 -0400

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6	pubmed:36119932	VEGFA-Enriched Exosomes from Tendon-Derived Stem Cells Facilitate Tenocyte Differentiation, Migration, and Transition to a Fibroblastic Phenotype	Zhaowen Xue Zihang Chen Tingting Wu Riwang Li Chao Chen Junting Liu Huige Hou Xiaofei Zheng Huajun Wang	Tendon-derived stem cells (TDSCs) play a vital role in repair of rotator cuff tear injuries by secreting paracrine proteins that regulate resident cell functions. Secreted exosomes may play a role in tendon injury repair by mediating intercellular communication; however, the detailed mechanisms by which TDSC-derived exosomes affect tenocyte development remain unknown. Here, we examined the effects of exosomes isolated from conditioned medium of TDSCs on tenocyte differentiation, migration, and...	pmid:36119932 pmc:PMC9481323 doi:10.1155/2022/8537959	Mon, 19 Sep 2022 06:00:00 -0400
7	pubmed:36120427	Coordinated regulation of gene expression and microRNA changes in adipose tissue and circulating extracellular vesicles in response to pioglitazone treatment in humans with type 2 diabetes	Yury O Nunez Lopez Anna Casu Zuzana Kovacova Alejandra M Petrilli Olga Sideleva William G Tharp Richard E Pratley	Pioglitazone, a PPAR agonist, is used to treat type 2 diabetes (T2D). PPAR is highly expressed in adipose tissue (AT), however the effects of pioglitazone to improve insulin sensitivity are also evident in other tissues and PPAR agonism has been shown to alter cancer derived extracellular vesicle (EV)-miRNAs. We hypothesized that pioglitazone modifies the cargo of circulating AT-derived EVs to alter interorgan crosstalk in people with diabetes. We tested our hypothesis in a 3-month trial in...	pmid:36120427 pmc:PMC9471675 doi:10.3389/fendo.2022.955593	Mon, 19 Sep 2022 06:00:00 -0400
8	pubmed:36120434	The miR-33a-5p/CROT axis mediates ovarian cancer cell behaviors and chemoresistance *via* the regulation of the TGF- signal pathway	Xin Li Xuzhu Gao Jia Yuan Fancheng Wang Xiaolin Xu Chenglong Wang Huiqiang Liu Wencai Guan Jihong Zhang Guoxiong Xu	Due to the lack of symptoms and detection biomarkers at the early stage, most patients with ovarian cancer (OC) are diagnosed at an advanced stage and often face chemoresistance and relapse. Hence, defining detection biomarkers and mechanisms of chemoresistance is imperative. A previous report of a cDNA microarray analysis shows a potential association of carnitine O-octanoyltransferase (CROT) with taxane resistance but the biological function of CROT in OC remains unknown. The current study...	pmid:36120434 pmc:PMC9478117 doi:10.3389/fendo.2022.950345	Mon, 19 Sep 2022 06:00:00 -0400
9	pubmed:36122615	Anti-metastatic breast cancer potential of novel nanocomplexes of diethyldithiocarbamate and green chemically synthesized iron oxide nanoparticles	Marwa M Abu-Serie Eisayeda Zeinab A Abdelfattah	Mortality rate of metastatic breast cancer is linked to cancer stem cells (CSCs)' aggressive features (chemoresistance to apoptosis and redox imbalance). Therefore, unique dual therapeutic strategy compacts CSCs with inducing oxidative stress-mediated nonapoptosis (ferroptosis), confers effective malignant tumor eradication. Diethyldithiocarbamate (DDC) is a potent inhibitor of CSC aldehyde dehydrogenase and lowers glutathione (GSH) which aggravate iron-dependent ferroptosis. Herein,...	pmid:36122615 doi:10.1016/j.ijpharm.2022.122208	Mon, 19 Sep 2022 06:00:00 -0400
10	pubmed:36122770	LncRNA Miat promotes neuropathic pain through miR-362-3p/BAMBI signaling axis	Wanyun Zhang Liming Zhou Chen Zhang	The treatment of neuropathic pain (NP) has become an important subject to be studied and solved urgently in clinical practice. The role of long noncoding RNAs (lncRNAs) in NP development is becoming clear. Therefore, this study aimed to investigate the role and mechanism of lncRNA Miat in NP. In this study, chronic contractionary injury (CCI) mouse NP model was performed. Firstly, the effects of Miat on pain behavior in mice and the expression levels of pro-inflammatory cytokines and...	pmid:36122770 doi:10.1016/j.yexcr.2022.113359	Mon, 19 Sep 2022 06:00:00 -0400

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11	pubmed:36122781	A concise review of VEGF, PDGF, FGF, Notch, angiopoietin, and HGF signalling in tumor angiogenesis with a focus on alternative approaches and future directions	Selvaraj Vimalraj	Angiogenesis forms new vessels from existing ones. Abnormal angiogenesis, which is what gives tumor microenvironments their distinctive features, is characterised by convoluted, permeable blood vessels with a variety of shapes and high perfusion efficiency. Tumor angiogenesis controls cancer growth by allowing invasion and metastasis and is highly controlled by signalling networks. Therapeutic techniques targeting VEGF, PDGF, FGF Notch, Angiopoietin, and HGF signalling restrict the tumor's...	pmid:36122781 doi:10.1016/j.ijbiomac.2022.09.129	Mon, 19 Sep 2022 06:00:00 -0400
12	pubmed:36122795	A novel prognostic index of stomach adenocarcinoma based on immunogenomic landscape analysis and immunotherapy options	Weijie Xue Bingzi Dong Yixiu Wang Yuwei Xie Pu Li Zhiqi Gong Zhaojian Niu	Stomach adenocarcinoma (STAD) is one of the most common malignant tumors worldwide. In this study, we attempted to construct a valid immune-associated gene prognostic index risk model that can predict the survival of patients with STAD and the efficacy of immune checkpoint inhibitors (ICIs) treatment. Transcriptome, clinical, and gene mutational data were obtained from the TCGA database. Immune-related genes were downloaded from the ImmPort and InnateDB databases. A total of 493 immune-related...	pmid:36122795 doi:10.1016/j.yexmp.2022.104832	Mon, 19 Sep 2022 06:00:00 -0400
13	pubmed:36122928	Development of a New DHFR-Based Destabilizing Domain with Enhanced Basal Turnover and Applicability in Mammalian Systems	Emi Nakahara Vishruth Mullapudi Gracen E Collier Lukasz A Joachimiak John D Hulleman	Destabilizing domains (DDs) are an attractive strategy allowing for positive post-transcriptional small molecule-regulatable control of a fusion protein's abundance. However, in many instances, the currently available DDs suffer from higher-than-desirable basal levels of the fusion protein. Accordingly, we redesigned the E. coli dihydrofolate reductase (ecDHFR) DD by introducing a library of 1200 random ecDHFR mutants fused to YFP into CHO cells. Following successive rounds of...	pmid:36122928 doi:10.1021/acschembio.2c00518	Mon, 19 Sep 2022 06:00:00 -0400
14	pubmed:36122933	Novel Lymphocytic Choriomeningitis Virus Strain Sustains Abundant Exhausted Progenitor CD8 T Cells without Systemic Viremia	Lalit K Beura Milcah C Scott Mark J Pierson Vineet Joag Sathi Wijeyesinghe Matthew R Semler Clare F Quarnstrom Kathleen Busman-Sahay Jacob D Estes Sara E Hamilton Vaiva Vezys David H O'Connor David Masopust	Lymphocytic choriomeningitis virus (LCMV) is the prototypic arenavirus and a natural mouse pathogen. LCMV-Armstrong, an acutely resolved strain, and LCMV-clone 13, a mutant that establishes chronic infection, have provided contrasting infection models that continue to inform the fundamental biology of T cell differentiation, regulation of exhaustion, and response to checkpoint blockade. In this study, we report the isolation and characterization of LCMV-Minnesota (LCMV-MN), which was naturally...	pmid:36122933 doi:10.4049/jimmunol.2200320	Mon, 19 Sep 2022 06:00:00 -0400
15	pubmed:36123150	Therapeutic Approaches Targeting miRNA in Systemic Lupus Erythematosus	Sumie Hiramatsu-Asano Jun Wada	Systemic lupus erythematosus (SLE) is a potentially fatal systemic autoimmune disease, and its etiology involves both genetic and environmental factors such as sex hormone imbalance, genetic predisposition, epigenetic regulation, and immunological factors. Dysregulation of microRNA (miRNA) is suggested to be one of the epigenetic factors in SLE. miRNA is a 22-nucleotide single-stranded noncoding RNA that contributes to post-transcriptional modulation of gene expression. miRNA targeting therapy...	pmid:36123150 doi:10.18926/AMO/63887	Mon, 19 Sep 2022 06:00:00 -0400

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16	pubmed:36123154	Gene Expression Profiling between Patient Groups with High and Low Ki67 Levels after Short-term Preoperative Aromatase Inhibitor Treatment for Breast Cancer	Yukiko Kajiwara Takayuki Iwamoto Yidan Zhu Mariko Kochi Tadahiko Shien Naruto Taira Hiroyoshi Doihara Shinichi Toyooka	According to a recent report, a low Ki67 level after short-term preoperative hormone therapy (post-Ki67) might suggest a more favorable prognosis compared with a high post-Ki67 level in patients with hormone receptorpositive/human epidermal growth factor 2-negative (HR+/HER2-) breast cancer with high levels of Ki67. This study aimed to evaluate the pre-treatment genetic differences between these two patient groups. Forty-five luminal B-like patients were stratified into two groups, namely, a...	pmid:36123154 doi:10.18926/AMO/63894	Mon, 19 Sep 2022 06:00:00 -0400
17	pubmed:36123222	Postoperative day one systemic inflammatory response syndrome is a powerful early biomarker of clinically relevant pancreatic fistula	F Ausania C Gonzalez-Abós A Martinez-Perez C Arrocha C Pineda-Garcés F Landi C Fillat J C Garcia-Valdecasas	CONCLUSION: POD1 SIRS is powerfully associated with CR-POPF and therefore it could be used as a tool to optimize postoperative care of PD patients. Further prospective studies are needed to validate these findings.	pmid:36123222 doi:10.1016/j.hpb.2022.08.016	Mon, 19 Sep 2022 06:00:00 -0400
18	pubmed:36123234	Editing human hematopoietic stem cells: advances and challenges	Senthil Velan Bhoopalan Jonathan S Yen Rachel M Levine Akshay Sharma	Genome editing of hematopoietic stem and progenitor cells is being developed for the treatment of several inherited disorders of the hematopoietic system. The adaptation of CRISPR-Cas9-based technologies to make precise changes to the genome, and developments in altering the specificity and efficiency, and improving the delivery of nucleases to target cells have led to several breakthroughs. Many clinical trials are ongoing, and several pre-clinical models have been reported that would allow...	pmid:36123234 doi:10.1016/j.jcyt.2022.08.003	Mon, 19 Sep 2022 06:00:00 -0400
19	pubmed:36123393	Single nuclei transcriptomics of muscle reveals intra-muscular cell dynamics linked to dystrophin loss and rescue	Deirdre D Scripture-Adams Kevin N Chesmore Florian Barthélémy Richard T Wang Shirley Nieves-Rodriguez Derek W Wang Ekaterina I Mokhonova Emilie D Douine Jijun Wan Isaiah Little Laura N Rabichow Stanley F Nelson M Carrie Miceli	In Duchenne muscular dystrophy, dystrophin loss leads to chronic muscle damage, dysregulation of repair, fibro-fatty replacement, and weakness. We develop methodology to efficiently isolate individual nuclei from minute quantities of frozen skeletal muscle, allowing single nuclei sequencing of irreplaceable archival samples and from very small samples. We apply this method to identify cell and gene expression dynamics within human DMD and mdx mouse muscle, characterizing effects of dystrophin...	pmid:36123393 doi:10.1038/s42003-022-03938-0	Mon, 19 Sep 2022 06:00:00 -0400
20	pubmed:36123426	Prospective observational comparison of arteria uterina blood flow between two frozen embryo transfer cycle regimens: natural cycle versus hormonal replacement cycle	Barbara Lawrenz Desislava Markova Laura Melado Raquel Loja Vitorino Shieryl Digma Suzan Samir Human M Fatemi	CONCLUSIONS: Early measurements of the blood flow parameters during the FET cycle do not reveal a difference between the NC- and the HRT-approach for FET, which could be predictive for development of pre-eclampsia.	pmid:36123426 doi:10.1007/s00404-022-06789-6	Mon, 19 Sep 2022 06:00:00 -0400

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21	pubmed:36123612	Immune dysregulation associated with co-occurring germline CBL and SH2B3 variants	Francesco Baccelli Davide Leardini Edoardo Muratore Daria Messelodi Salvatore Nicola Bertuccio Maria Chiriaco Caterina Cancrini Francesca Conti Fausto Castagnetti Lucia Pedace Andrea Pession Ayami Yoshimi Charlotte Niemeyer Marco Tartaglia Franco Locatelli Riccardo Masetti	CONCLUSION: In the reported family, we described immune dysregulation, as part of the clinical spectrum of CBL mutation with the co-occurrence of SH2B3.	pmid:36123612 doi:10.1186/s40246-022-00414-y	Mon, 19 Sep 2022 06:00:00 -0400
22	pubmed:36123640	Clinical and genetic factors associated with self-reported cognitive deficits in women with breast cancer: the "CAGE-Cog" study	Aline Hajj Rita Khoury Roula Hachem Aya Awad Souheil Hallit Hala Sacre Fady Nasr Fadi El Karak Georges Chahine Joseph Kattan Lydia Rabbaa Khabbaz	CONCLUSIONS: A comprehensive oncological care plan should include a personalized assessment of all factors related to cognitive functioning in cancer patients, particularly anxiety and depression, to achieve an optimal patient outcome.	pmid:36123640 doi:10.1186/s12885-022-10077-6	Mon, 19 Sep 2022 06:00:00 -0400
23	pubmed:36123698	Transcription factor AP2 enhances malignancy of non-small cell lung cancer through upregulation of USP22 gene expression	Ting Sun Keqiang Zhang Wendong Li Yunze Liu Rajendra P Pangeni Aimin Li Leonidas Arvanitis Dan J Raz	CONCLUSION: Our findings indicate AP2 and AP2 are important transcription factors driving USP22 gene expression to promote the progression of NSCLC, and further support USP22 as a potential biomarker and therapeutic target for lung cancer. Video Abstract.	pmid:36123698 doi:10.1186/s12964-022-00946-9	Mon, 19 Sep 2022 06:00:00 -0400
24	pubmed:36123711	The immune microenvironment of HPV-positive and HPV-negative oropharyngeal squamous cell carcinoma: a multiparametric quantitative and spatial analysis unveils a rationale to target treatment-naïve tumors with immune checkpoint inhibitors	Anna Tosi Beatrice Parisatto Anna Menegaldo Giacomo Spinato Maria Guido Annarosa Del Mistro Rossana Bussani Fabrizio Zanconati Margherita Tofanelli Giancarlo Tirelli Paolo Boscolo-Rizzo Antonio Rosato	CONCLUSIONS: Our results suggest that checkpoint expression may reflect an ongoing antitumor immune response. Thus, these observations provide the rationale for the incorporation of ICI in the loco-regional therapy strategies for patients with heavily infiltrated treatment-naïve OPSCC, and for the combination of ICI with tumor-specific T cell response inducers or TAM modulators for the "cold" OPSCC counterparts.	pmid:36123711 doi:10.1186/s13046-022-02481-4	Mon, 19 Sep 2022 06:00:00 -0400
25	pubmed:36123877	Prognostic value of m6A regulators and the nomogram construction in glioma patients	Pengdi Liu Xianxia Yan Chengwen Ma Junxiang Gu Fuyu Tian Jianqiang Qu	Although N6-methyladenosine (m6A) has been implicated in various biological functions in human cancers, its role in predicting the prognosis of glioma remains unclear. In this study, the transcriptome expression profiles and the clinical data of 961 patients were derived from the Chinese Glioma Genome Atlas (CGGA). We comprehensively evaluated the association between the expression of m6A regulators and the prognosis of glioma and established a 3-gene (YTHDF2, FTO, and ALKBH5) risk signature...	pmid:36123877 doi:10.1097/MD.0000000000030643	Tue, 20 Sep 2022 06:00:00 -0400

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26	pubmed:36123899	Hub genes for early diagnosis and therapy of adamantinomatous craniopharyngioma	Yang-Fan Zou Shu-Yuan Zhang Li-Weng Li Kai Jing Liang Xia Cai-Xing Sun Bin Wu	CONCLUSION: In a word, we find out the DEGs between ACP patients and standard samples, which are likely to play an essential role in the development of ACP. At the same time, these DEGs are of great value in tumors' diagnosis and targeted therapy and could even be mined as biological molecular targets for diagnosing and treating ACP patients.	pmid:36123899 doi:10.1097/MD.0000000000030278	Tue, 20 Sep 2022 06:00:00 -0400
27	pubmed:36124037	Multidisciplinary Approach to Treating Chronic Pain in Patients with Ehlers-Danlos Syndrome: Critically Appraised Topic	Kiley C Whalen Wilson Crone	The Ehlers-Danlos syndromes (EDS) are a group of 13 related connective tissue disorders with a combined prevalence of 1 in 5000 people, with the most common noted as hypermobile EDS (hEDS). The EDS genetic condition is thought to affect both males and females equally, although most symptomatic patients are female. EDS causes a myriad of symptoms, including skin hyperextensibility and fragility, easy bruising and bleeding, joint hypermobility, subluxation, dislocation, and chronic pain. Pain is...	pmid:36124037 pmc:PMC9482467 doi:10.2147/JPR.S377790	Tue, 20 Sep 2022 06:00:00 -0400
28	pubmed:36124066	Effects of CoQ10 Replacement Therapy on the Audiological Characteristics of Pediatric Patients with *COQ6* Variants	Dong Woo Nam Sang Soo Park So Min Lee Myung-Whan Suh Moo Kyun Park Jae-Jin Song Byung Yoon Choi Jun Ho Lee Seung Ha Oh Kyung Chul Moon Yo Han Ahn Hee Gyung Kang Hae Il Cheong Ji Hyun Kim Sang-Yeon Lee	Primary coenzyme Q10 (CoQ10) deficiency refers to a group of mitochondrial cytopathies caused by genetic defects in CoQ10 biosynthesis. Primary coenzyme Q10 deficiency-6 (COQ10D6) is an autosomal recessive disorder attributable to biallelic COQ6 variants; the cardinal phenotypes are steroid-resistant nephrotic syndrome (SRNS), which inevitably progresses to kidney failure, and sensorineural hearing loss (SNHL). Here, we describe the phenotypes and genotypes of 12 children with COQ10D6 from 11...	pmid:36124066 pmc:PMC9482153 doi:10.1155/2022/5250254	Tue, 20 Sep 2022 06:00:00 -0400
29	pubmed:36124139	Exploration of Hub Genes in Retinopathy of Prematurity Based on Bioinformatics Analysis of the Oxygen-Induced Retinopathy Model	Qi Xiong Zhiliang Li Jing Zhang Lin Yang Xiaomin Chen Yan Gong Xiaojun Cai Min Ke	Retinopathy of prematurity (ROP) is a major blindness-causing disease that is characterized by an arrest of normal vascular development and neovascularization of the retina. Previous studies have shown that genetic factors may be associated with the development and severity of ROP. However, the genes and mechanisms underlying ROP remain unclear. We aimed to identify hub genes in ROP and drugs related to these genes by integrative analysis. The expression profiles of GSE158799 and GSE135844 were...	pmid:36124139 pmc:PMC9482502 doi:10.1155/2022/9835524	Tue, 20 Sep 2022 06:00:00 -0400
30	pubmed:36124593	Development and validation of novel inflammatory response-related gene signature to predict prostate cancer recurrence and response to immune checkpoint therapy	Yong Luo Xiaopeng Liu Jingbo Lin Weide Zhong Qingbiao Chen	The aim of this study is to construct an inflammatory response-related genes (IRRGs) signature to monitor biochemical recurrence (BCR) and treatment effects in prostate cancer patients (PCa). A gene signature for inflammatory responses was constructed on the basis of the data from the Cancer Genome Atlas (TCGA) database, and validated in external datasets. It was analyzed using receiver operating characteristic curve, BCR-free survival, Cox regression, and nomogram. Distribution analysis and...	pmid:36124593 doi:10.3934/mbe.2022528	Tue, 20 Sep 2022 06:00:00 -0400

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31	pubmed:36124682	Super-enhancer-controlled positive feedback loop BRD4/ER-RET-ER promotes ER-positive breast cancer	Zao-Zao Zheng Lin Xia Guo-Sheng Hu Jun-Yi Liu Ya-Hong Hu Yu-Jie Chen Jia-Yin Peng Wen-Juan Zhang Wen Liu	Estrogen and estrogen receptor alpha (ER)-induced gene transcription is tightly associated with ER-positive breast carcinogenesis. ER-occupied enhancers, particularly super-enhancers, have been suggested to play a vital role in regulating such transcriptional events. However, the landscape of ER-occupied super-enhancers (ERSEs) as well as key ER-induced target genes associated with ERSEs remain to be fully characterized. Here, we defined the landscape of ERSEs in ER-positive breast cancer...	pmid:36124682 doi:10.1093/nar/gkac778	Tue, 20 Sep 2022 06:00:00 -0400
32	pubmed:36124778	Current and promising therapeutic options for Dravet Syndrome	Antonella Riva Gianluca D'Onofrio Elisabetta Amadori Domenico Tripodi Ganna Balagura Valentina Iurilli Maria Stella Vari Alberto Verrotti Pasquale Striano	INTRODUCTION: Dravet Syndrome (DS) is a developmental and epileptic encephalopathy carrying high-level psychobehavioral burdens. Although the disease has been known for almost 4 decades, and despite significant progress in the understanding of its physiopathology and natural course, the pharmacological treatment leaves patients and caregivers with significant unmet needs. This review provides a summary of the current and promising therapeutic options for DS.	pmid:36124778 doi:10.1080/14656566.2022.2127089	Tue, 20 Sep 2022 06:00:00 -0400
33	pubmed:36124781	An evaluation of mitapivat for the treatment of hemolytic anemia in adults with pyruvate kinase deficiency	Andrew B Song Hanny Al-Samkari	INTRODUCTION: Pyruvate kinase deficiency (PKD) is the most common cause of congenital nonspherocytic hemolytic anemia. Until recently, treatment had been limited to supportive management including red blood cell transfusions, splenectomy, and management of chronic disease complications such as iron overload and decreased bone mineral density.	pmid:36124781 doi:10.1080/17474086.2022.2125865	Tue, 20 Sep 2022 06:00:00 -0400
34	pubmed:36124999	Inhibition of O-GlcNAcase inhibits hematopoietic and leukemic stem cell self-renewal and drives dendritic cell differentiation via STAT3/5 signaling	Sudjit Luanpitpong Napachai Rodboon Parinya Samart Montira Janan Phatchanat Klaihmon Chanchao Lorthongpanich Yaowalak U-Pratya Surapol Issaragrisil	Myeloid differentiation blockage at immature and self-renewing stages is a common hallmark across all subtypes of acute myeloid leukemia (AML), despite their genetic heterogeneity. Metabolic state is an important regulator of hematopoietic stem cell (HSC) self-renewal and lineage-specific differentiation as well as several aggressive cancers. However, how O-GlcNAcylation, a nutrient-sensitive posttranslational modification of proteins, contributes to both normal myelopoiesis and AML pathogenesis...	pmid:36124999 doi:10.1093/stmcls/sxac068	Tue, 20 Sep 2022 06:00:00 -0400
35	pubmed:36125010	Novel pathophysiological roles of -synuclein in age-related vascular endothelial dysfunction	Yoichi Takami Cheng Wang Hironori Nakagami Koichi Yamamoto Yoichi Nozato Yuki Imaizumi Motonori Nagasawa Hikari Takeshita Tsuneo Nakajima Shuko Takeda Yasushi Takeya Yasufumi Kaneda Hiromi Rakugi	Although -synuclein (SNCA) is a well-known pathological molecule involved in synucleinopathy in neurons, its physiological roles remain largely unknown. We reported that serum SNCA levels have a close inverse correlation with blood pressure and age, which indicates the involvement of SNCA in age-related endothelial dysfunction. Therefore, this study aimed to elucidate the molecular functions of SNCA in the endothelium. We confirmed that SNCA was expressed in and secreted from endothelial cells...	pmid:36125010 doi:10.1096/fj.202101621R	Tue, 20 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
36	pubmed:36125035	Management of front line chronic lymphocytic leukemia	Nadine Kutsch Anna Maria Fink Kirsten Fischer	Treatment options with targeted agents have changed the treatment landscape of CLL profoundly. Besides chemoimmunotherapy, treatment regimen approved for frontline therapy include continuous treatment with BTK inhibitors like ibrutinib and acalabrutinib or fixed-duration regimen like venetoclax-obinutuzumab with the approval of venetoclax-ibrutinib to be awaited. Although these agents have usually manageable side effects, toxicities might limit choices for the individual patient. We here discuss...	pmid:36125035 doi:10.1002/ajh.26677	Tue, 20 Sep 2022 06:00:00 -0400
37	pubmed:36125320	Dual enzyme therapy improves adherence to chemotherapy in a patient with gaucher disease and Ewing sarcoma	Brandon Lucari Eran Tallis Vernon Reid Sutton Timothy Porea	This case reports concomitant use of enzyme and substrate reduction therapy to improve chemotherapy adherence in a pediatric patient diagnosed with Ewing sarcoma (ES) and type 1 Gaucher disease (GD). The 17-year-old female presented with 5 months of right knee pain with associated mass on exam. She was diagnosed with ES with pulmonary metastasis. The patient was treated with 17 alternating cycles of vincristine-doxorubicin-cyclophosphamide and ifosfamide and etoposide chemotherapy followed by...	pmid:36125320 doi:10.1080/08880018.2022.2124006	Tue, 20 Sep 2022 06:00:00 -0400
38	pubmed:36125329	Thymosin 4 and prothymosin promote cardiac regeneration post-ischemic injury in mice	Monika M Gladka Anne Katrine Z Johansen Sebastiaan J van Kampen Marijn M C Peters Bas Molenaar Danielle Versteeg Lieneke Kooijman Lorena Zentilin Mauro Giacca Eva van Rooij	The adult mammalian heart is a post-mitotic organ. Even in response to necrotic injuries, where regeneration would be essential to reinstate cardiac structure and function, only a minor percentage of cardiomyocytes undergo cytokinesis. The gene program that promotes cell division within this population is not fully understood. Here, we demonstrate increased EdU incorporation in cardiomyocytes at 3 days post-myocardial infarction (MI) in mice. By applying multi-color lineage tracing, we show that...	pmid:36125329 doi:10.1093/cvr/cvac155	Tue, 20 Sep 2022 06:00:00 -0400
39	pubmed:36125338	mRNA Treatment Rescues Niemann-Pick Disease Type C1 in Patient Fibroblasts	Denzil Furtado Christina Cortez-Jugo Ya Hui Hung Ashley I Bush Frank Caruso	Messenger RNA (mRNA) holds great potential as a disease-modifying treatment for a wide array of monogenic disorders. Niemann-Pick disease type C1 (NP-C1) is an ultrarare monogenic disease that arises due to loss-of-function mutations in the NPC1 gene, resulting in the entrapment of unesterified cholesterol in the lysosomes of affected cells and a subsequent reduction in their capacity for cholesterol esterification. This causes severe damage to various organs including the brain, liver, and...	pmid:36125338 doi:10.1021/acs.molpharmaceut.2c00463	Tue, 20 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
40	pubmed:36125345	Conversion therapy for initially unresectable hepatocellular carcinoma using a combination of toripalimab, lenvatinib plus TACE: real-world study	Wei-Feng Qu Zhen-Bin Ding Xu-Dong Qu Zheng Tang Gui-Qi Zhu Xiu-Tao Fu Zi-Han Zhang Xin Zhang Ao Huang Min Tang Meng-Xin Tian Xi-Fei Jiang Run Huang Chen-Yang Tao Yuan Fang Jun Gao Xiao-Ling Wu Jian Zhou Jia Fan Wei-Ren Liu Ying-Hong Shi	CONCLUSIONS: Better treatment responses and conversion rate for patients with uHCC were obtained with first-line t-CT. Neoadjuvant t-CT before surgery should be recommended for patients with macrovascular invasion.	pmid:36125345 doi:10.1093/bjsopen/zrac114	Tue, 20 Sep 2022 06:00:00 -0400
41	pubmed:36125365	Designing Synthetic Polymers for Nucleic Acid Complexation and Delivery: From Polyplexes to Micelleplexes to Triggered Degradation	Le Zhou Miracle Emenuga Shreya Kumar Zachary Lamantia Marxa Figueiredo Todd Emrick	Gene delivery as a therapeutic tool continues to advance toward impacting human health, with several gene therapy products receiving FDA approval over the past 5 years. Despite this important progress, the safety and efficacy of gene therapy methodology requires further improvement to ensure that nucleic acid therapeutics reach the desired targets while minimizing adverse effects. Synthetic polymers offer several enticing features as nucleic acid delivery vectors due to their versatile...	pmid:36125365 doi:10.1021/acs.biomac.2c00767	Tue, 20 Sep 2022 06:00:00 -0400
42	pubmed:36125411	Retinal and Choroidal Neovascularization Antivascular Endothelial Growth Factor Treatments: The Role of Gene Therapy	Samaneh Toutounchian Naser Ahmadbeigi Vahid Mansouri	Neovascularization in ocular vessels causes a major disease burden. The most common causes of choroidal neovascularization (CNV) are age-related macular degeneration and diabetic retinopathy, which are the leading causes of irreversible vision loss in the adult population. Vascular endothelial growth factor (VEGF) is critical for the formation of new vessels and is the main regulator in ocular angiogenesis and vascular permeability through its receptors. Laser therapy and antiangiogenic factors...	pmid:36125411 doi:10.1089/jop.2022.0022	Tue, 20 Sep 2022 06:00:00 -0400
43	pubmed:36125439	Novartis Confirms Deaths of Two Patients Treated with Gene Therapy Zolgensma	Alex Philippidis	No abstract	pmid:36125439 doi:10.1089/hum.2022.29216.bfs	Tue, 20 Sep 2022 06:00:00 -0400
44	pubmed:36125440	ESGCT 2022: The New Normal for the Gene Therapy Community	Terence R Flotte	No abstract	pmid:36125440 doi:10.1089/hum.2022.29217.editorial	Tue, 20 Sep 2022 06:00:00 -0400
45	pubmed:36125483	New Gene Therapy for -Thalassemia	Rita Rubin	No abstract	pmid:36125483 doi:10.1001/jama.2022.14709	Tue, 20 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
46	pubmed:36125660	Molecular characterization of ESR1 variants in breast cancer	Arielle L Heeke Andrew Elliott Rebecca Feldman Hazel F O'Connor Paula R Pohlmann Filipa Lynce Sandra M Swain Maria R Nunes Daniel Magee Matthew J Oberley Jeffrey Swenson Gregory Vidal Claudine Isaacs Lee Schwartzberg W Michael Korn Antoinette R Tan	CONCLUSION: We have described one of the largest series of ESR1 fusions reported. ESR1 LBD mutations were commonly identified in ER-positive disease. Limited data exists regarding the clinical impact of ESR1 fusions, which could be an area for future therapeutic exploration.	pmid:36125660 doi:10.1007/s10549-022-06740-y	Tue, 20 Sep 2022 06:00:00 -0400
47	pubmed:36125711	Human Hepatocyte Transduction with Adeno-Associated Virus Vector	Zhenwei Song Wenwei Shao Liujiang Song Xieolei Pei Chengwen Li	As the adeno-associated virus (AAV) vectors hold unique advantages over other viral vectors, AAV gene therapy has accumulated rapid progress and development. Liver-targeted gene therapy by AAV vectors has been successfully applied in clinical trials for many diseases. Low transduction efficiency and high prevalence of neutralizing antibodies (Nabs), however, are the major obstacles to further translate this therapeutic strategy into clinical trials. Pre-clinical evaluation on hepatocytes could...	pmid:36125711 doi:10.1007/978-1-0716-2557-6_5	Tue, 20 Sep 2022 06:00:00 -0400
48	pubmed:36125723	Identification of Genes Regulating Hepatocyte Injury by a Genome-Wide CRISPR-Cas9 Screen	Katherine Shortt Daniel P Heruth	Gene editing introduces stable mutations into the genome and has powerful applications extending from research to clinical gene therapy. CRISPR-Cas9 gene editing can be employed to study directly the functional impact of stable gene knockout, activation, and knockdown. Here, we describe the end-to-end methodology by which we employ genome-wide CRISPR-Cas9 knockout to study drug toxicity using acetaminophen (APAP) in a hepatocellular carcinoma liver model as an example. This methodology can be...	pmid:36125723 doi:10.1007/978-1-0716-2557-6_17	Tue, 20 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
49	pubmed:36125896	H3.3-G34 mutations impair DNA repair and promote cGAS/STING-mediated immune responses in pediatric high-grade glioma models	Santiago Haase Kaushik Banerjee Anzar A Mujeeb Carson S Hartlage Fernando M Nunez Felipe J Nuñez Mahmoud S Alghamri Padma Kadiyala Stephen Carney Marcus Barissi Ayman W Taher Emily K Brumley Sarah Thompson Justin T Dreyer Caitlin T Alindogan Maria B Garcia-Fabiani Andrea Comba Sriram Venneti Visweswaran Ravikumar Carl Koschmann Angel M Carcaboso Maria Vinci Arvind Rao Jennifer S Yu Pedro R Lowenstein Maria G Castro	Pediatric high-grade gliomas (pHGGs) are the leading cause of cancer-related deaths in children in the USA. Sixteen percent of hemispheric pediatric and young adult HGGs encode Gly34Arg/Val substitutions in the histone H3.3 (H3.3-G34R/V). The mechanisms by which H3.3-G34R/V drive malignancy and therapeutic resistance in pHGGs remain unknown. Using a syngeneic, genetically engineered mouse model (GEMM) and human pHGG cells encoding H3.3-G34R, we demonstrate that this mutation leads to...	pmid:36125896 doi:10.1172/JCI154229	Tue, 20 Sep 2022 06:00:00 -0400
50	pubmed:36126056	Novel AAV-mediated genome editing therapy improves health and survival in a mouse model of methylmalonic acidemia	Shengwen Zhang Amy Bastille Susana Gordo Nikhil Ramesh Jenisha Vora Elizabeth McCarthy Xiaohan Zhang Dylan Frank Chih-Wei Ko Carmen Wu Noel Walsh Shreya Amarwani Jing Liao Qiang Xiong Lauren Drouin Matthias Hebben Kyle Chiang B Nelson Chau	Methylmalonic acidemia (MMA) is an inborn error of metabolism mostly caused by mutations in the mitochondrial methylmalonyl-CoA mutase gene (MMUT). MMA patients suffer from frequent episodes of metabolic decompensation, which can be life threatening. To mimic both the dietary restrictions and metabolic decompensation seen in MMA patients, we developed a novel protein-controlled diet regimen in a Mmut deficient mouse model of MMA and demonstrated the therapeutic benefit of mLB-001, a...	pmid:36126056 doi:10.1371/journal.pone.0274774	Tue, 20 Sep 2022 06:00:00 -0400
51	pubmed:36126293	Electrical, Electromagnetic, Ultrasound Wave Therapies, and Electronic Implants for Neuronal Rejuvenation, Neuroprotection, Axonal Regeneration, and IOP Reduction	Najam A Sharif	The peripheral nervous system (PNS) of mammals and nervous systems of lower organisms possess significant regenerative potential. In contrast, although neural plasticity can provide some compensation, the central nervous system (CNS) neurons and nerves of adult mammals generally fail to regenerate after an injury or damage. However, use of diverse electrical, electromagnetic and sonographic energy waves are illuminating novel ways to stimulate neuronal differentiation, proliferation, neurite...	pmid:36126293 doi:10.1089/jop.2022.0046	Tue, 20 Sep 2022 06:00:00 -0400