gene therapy

	NCT Number	Title	Authors	Description	Identifier	Dates
1	pubmed:36088683	Long-term antiretroviral therapy initiated in acute HIV infection prevents residual dysfunction of HIV-specific CD8^{+} T cells	Hiroshi Takata Juyeon C Kakazu Julie L Mitchell Eugene Kroon Donn J Colby Carlo Sacdalan Hongjun Bai Philip K Ehrenberg Aviva Geretz Supranee Buranapraditkun Suteeraporn Pinyakorn Jintana Intasan Somporn Tipsuk Duanghathai Suttichom Peeriya Prueksakaew Thep Chalermchai Nitiya Chomchey Nittaya Phanuphak Mark de Souza Nelson L Michael Merlin L Robb Elias K Haddad Trevor A Crowell Sandhya Vasan Victor G Valcour Daniel C Douek Rasmi Thomas Morgane Rolland Nicolas Chomont Jintanat Ananworanich Lydie Trautmann RV254/SEARCH010, RV304/ SEARCH013, and SEARCH011 study groups	BACKGROUND: Harnessing CD8^(+) T cell responses is being explored to achieve HIV remission. Although HIV-specific CD8^(+) T cells become dysfunctional without treatment, antiretroviral therapy (ART) partially restores their function. However, the extent of this recovery under long-term ART is less understood.	pmid:36088683 doi:10.1016/j.ebiom.2022.104253	Sun, 11 Sep 2022 06:00:00 -0400
2	pubmed:36088815	Cardiac involvement in Fabry disease - A non-invasive assessment and the role of specific therapies	Kenichi Hongo	Fabry disease is an X-linked inherited metabolic disorder due to the pathogenic mutation of the GLA gene, which codes lysosomal enzyme alpha-galactosidase A. The resultant accumulation of glycosphingolipids causes various systemic symptoms in childhood and adolescence, and major organ damage in adulthood. Cardiac involvement is important as the most frequent cause of death in Fabry disease patients. Progressive left ventricular hypertrophy with varying degrees of contractile dysfunction as well...	pmid:36088815 doi:10.1016/j.ymgme.2022.08.006	Sun, 11 Sep 2022 06:00:00 -0400
3	pubmed:36088816	Hermansky-Pudlak syndrome: Gene therapy for pulmonary fibrosis	Gustavo Nieto-Alamilla Molly Behan Mahin Hossain Bernadette R Gochuico May Christine V Malicdan	Pulmonary fibrosis is a progressive and often fatal lung disease that manifests in most patients with Hermansky-Pudlak syndrome (HPS) type 1. Although the pathobiology of HPS pulmonary fibrosis is unknown, several studies highlight the pathogenic roles of different cell types, including type 2 alveolar epithelial cells, alveolar macrophages, fibroblasts, myofibroblasts, and immune cells. Despite the identification of the HPS1 gene and progress in understanding the pathobiology of HPS pulmonary...	pmid:36088816 doi:10.1016/j.ymgme.2022.08.008	Sun, 11 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
4	pubmed:36088836	Effects of 5-ALA mediated photodynamic therapy in oral cancer stem cells	Marlene Aparecida Ferreira Pinto Cássia Bosi Ribeiro Ferreira Bárbara Evelyn Santos de Lima Ângela Cristina Molon Ana Melissa Coppa Ibarra Rebeca Boltes Cecatto Adriana Lino Dos Santos Franco Maria Fernanda Setúbal Destro Rodrigues	The aim of the present study was to investigate the effects of PDT using the photosensitizer 5-aminoulevulinic acid (5-ALA) in oral squamous cell carcinoma (OSCC) behavior, mainly regarding its role on the cancer stem cell (CSC) phenotypes and in maintenance of the stem cell properties. Two OSCC cell lines were used and divided in the groups: Control, 5-ALA, LED 6 J/cm² and PDT. MTT and Neutral red assays were used to access cellular viability, cell migration was evaluated by the wound healing...	pmid:36088836 doi:10.1016/j.jphotobiol.2022.112552	Sun, 11 Sep 2022 06:00:00 -0400
5	pubmed:36088954	COVID-19 vaccine-induced antibody and T-cell responses in immunosuppressed patients with inflammatory bowel disease after the third vaccine dose (VIP): a multicentre, prospective, case-control study	James L Alexander Zhigang Liu Diana Muñoz Sandoval Catherine Reynolds Hajir Ibraheim Sulak Anandabaskaran Aamir Saifuddin Rocio Castro Seoane Nikhil Anand Rachel Nice Claire Bewshea Andrea D'Mello Laura Constable Gareth R Jones Sharmili Balarajah Francesca Fiorentino Shaji Sebastian Peter M Irving Lucy C Hicks Horace R T Williams Alexandra J Kent Rachel Linger Miles Parkes Klaartje Kok Kamal V Patel Julian P Teare Daniel M Altmann James R Goodhand Ailsa L Hart Charlie W Lees Rosemary J Boyton Nicholas A Kennedy Tariq Ahmad Nick Powell VIP study investigators	BACKGROUND: COVID-19 vaccine-induced antibody responses are reduced in patients with inflammatory bowel disease (IBD) taking anti-TNF or tofacitinib after two vaccine doses. We sought to assess whether immunosuppressive treatments were associated with reduced antibody and T-cell responses in patients with IBD after a third vaccine dose.	pmid:36088954 doi:10.1016/S2468-1253(22)00274-6	Sun, 11 Sep 2022 06:00:00 -0400
6	pubmed:36089077	Control of cell metabolism by the epidermal growth factor receptor	Laura A Orofiamma Dafne Vural Costin N Antonescu	The epidermal growth factor receptor (EGFR) triggers the activation of many intracellular signals that control cell proliferation, growth, survival, migration, and differentiation. Given its wide expression, EGFR has many functions in development and tissue homeostasis. Some of the cellular outcomes of EGFR signaling involve alterations of specific aspects of cellular metabolism, and alterations of cell metabolism are emerging as driving influences in many physiological and pathophysiological...	pmid:36089077 doi:10.1016/j.bbamcr.2022.119359	Sun, 11 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
7	pubmed:36089134	APOBEC mutagenesis, kataegis, chromothripsis in EGFR-mutant osimertinib-resistant lung adenocarcinomas	P Selenica A Marra N J Choudhury A Gazzo C J Falcon J Patel X Pei Y Zhu C K Y Ng M Curry G Heller Y-K Zhang M F Berger M Ladanyi C M Rudin S Chandarlapaty C M Lovly J S Reis-Filho H A Yu	CONCLUSIONS: APOBEC mutational signatures are frequent in RTK-driven LUADs and increase under the selective pressure of osimertinib in EGFR-mutant lung cancer. APOBEC mutational signature enrichment in subclonal mutations, private mutations acquired after osimertinib treatment, and areas of large scale genomic rearrangements highlights a potentially fundamental role for APOBEC mutagenesis in the development of resistance to targeted therapies, which may be potentially exploited to overcome such...	pmid:36089134 doi:10.1016/j.annonc.2022.09.151	Sun, 11 Sep 2022 06:00:00 -0400
8	pubmed:36089135	The Clinical Landscape of Cell-Free DNA Alterations in 1,671 Patients with Advanced Biliary Tract Cancer	Jacob E Berchuck Francesco Facchinetti Daniel F DiToro Islam Baiev Umair Majeed Stephanie Reyes Christopher Chen Karen Zhang Reya Sharman Pedro Luiz Serrano Uson Junior Jordan Maurer Rachna T Shroff Colin C Pritchard Meng-Ju Wu Daniel V T Catenacci Milind Javle Luc Friboulet Antoine Hollebecque Nabeel Bardeesy Andrew X Zhu Jochen K Lennerz Benjamin Tan Mitesh Borad Aparna R Parikh Lesli A Kiedrowski Robin Kate Kelley Kabir Mody Dejan Juric Lipika Goyal	CONCLUSIONS: These findings from the largest and most comprehensive study to date of cfDNA from patients with advanced BTC highlight the utility of cfDNA analysis in current management of this disease. Characterization of oncogenic drivers and mechanisms of therapeutic resistance in this study will inform drug development efforts to reduce mortality for patients with BTC.	pmid:36089135 doi:10.1016/j.annonc.2022.09.150	Sun, 11 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
9	pubmed:36089156	Human duodenal submucosal glands contain a defined stem/progenitor subpopulation with liver-specific regenerative potential	Vincenzo Cardinale Guido Carpino Diletta Overi Samira Safarikia Wencheng Zhang Matt Kanke Antonio Franchitto Daniele Costantini Olga Riccioni Lorenzo Nevi Michele Chiappetta Paolo Onori Matteo Franchitto Simone Bini Yu-Han Hung Quirino Lai Ilaria Zizzari Marianna Nuti Carmine Nicoletti Saula Checquolo Laura Di Magno Maria Valeria Giuli Massimo Rossi Praveen Sethupathy Lola M Reid Domenico Alvaro Eugenio Gaudio	CONCLUSIONS: A cell population with clonal growth and organoid formation capability, and which has liver differentiation potency in vitro and in vivo in murine experimental models, is present within adult duodenal submucosal glands. These cells can be isolated, do not require reprogramming, and thus could potentially represent a novel cell source for regenerative medicine of the liver.	pmid:36089156 doi:10.1016/j.jhep.2022.08.037	Sun, 11 Sep 2022 06:00:00 -0400
10	pubmed:36089170	Prediction the clinical EPR effect of nanoparticles in patient-derived xenograft models	Sangmin Jeon Eunsung Jun Hyeyoun Chang Ji Young Yhee Eun-Young Koh Yeounhee Kim Jae Yun Jung Eun Ji Jeong Jong Won Lee Man Kyu Shim Hong Yeol Yoon Suhwan Chang Kwangmeyung Kim Song Cheol Kim	Many preclinically tested nanoparticles in existing animal models fail to be directly translated into clinical applications because of their poor resemblance to human cancer. Herein, the enhanced permeation and retention (EPR) effect of glycol chitosan nanoparticles (CNPs) in different tumor microenvironments (TMEs) was compared using different pancreatic tumor models, including pancreatic cancer cell line (BxPC3), patient-derived cancer cell (PDC), and patient-derived xenograft (PDX) models....	pmid:36089170 doi:10.1016/j.jconrel.2022.09.007	Sun, 11 Sep 2022 06:00:00 -0400
11	pubmed:36089182	Liposomal formulations for treating lysosomal storage disorders	Judit Tomsen-Melero Josep Merlo-Mas Aida Carreño Santi Sala Alba Córdoba Jaume Veciana Elisabet González-Mira Nora Ventosa	Lysosomal storage disorders (LSD) are a group of rare life-threatening diseases caused by a lysosomal dysfunction, usually due to the lack of a single enzyme required for the metabolism of macromolecules, which leads to a lysosomal accumulation of specific substrates, resulting in severe disease manifestations and early death. There is currently no definitive cure for LSD, and despite the approval of certain therapies, their effectiveness is limited. Therefore, an appropriate nanocarrier could...	pmid:36089182 doi:10.1016/j.addr.2022.114531	Sun, 11 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
12	pubmed:36089496	Medical treatment of patients with hypertrophic cardiomyopathy: An overview of current and emerging therapy	Michele Iavarone Emanuele Monda Olga Vritz Dimpna Calila Albert Marta Rubino Federica Verrillo Martina Caiazza Michele Lioncino Federica Amodio Natale Guarnaccia Felice Gragnano Raffaella Lombardi Giovanni Esposito Eduardo Bossone Paolo Calabrò Maria Angela Losi Giuseppe Limongelli	Several treatments have demonstrated safety and effectiveness in the treatment of patients with hypertrophic cardiomyopathy; however, no drug has been shown to modify the natural history of the disease or to decrease maximal wall thickness. Improvement in our knowledge of the physiopathology of the disease has permitted the development of new therapeutical approaches, including sarcomere modulators and gene therapy. A sarcomere modulator - mavacamten - has been shown to improve exercise...	pmid:36089496 doi:10.1016/j.acvd.2022.06.003	Sun, 11 Sep 2022 06:00:00 -0400
13	pubmed:36089506	Liquid biopsy for the detection of resistance mutations to ROS1 and RET inhibitors in non-small lung cancers: A case series study	Yoshitaka Seki Tatsuya Yoshida Takashi Kohno Ken Masuda Yusuke Okuma Yasushi Goto Hidehito Horinouchi Noboru Yamamoto Kazuyoshi Kuwano Yuichiro Ohe	Liquid biopsy can identify gene alterations that are associated with resistance to fusion gene-targeted treatments. In this study, we present three cases of advanced non-small cell lung cancer (NSCLC) harboring gene fusions; cell-free DNA (cfDNA) was used to assess the resistance mutations. A patient with MET amplification underwent RET-fusion NSCLC treatment with selpercatinib. A patient with ROS1 G2032R underwent ROS1-fusion NSCLC treatment with crizotinib. A patient who underwent ROS1-fusion...	pmid:36089506 doi:10.1016/j.resinv.2022.08.002	Sun, 11 Sep 2022 06:00:00 -0400
14	pubmed:36089578	Characterization of the T cell receptor repertoire and melanoma tumor microenvironment upon combined treatment with ipilimumab and hTERT vaccination	Espen Basmo Ellingsen Gergana Bounova Iliana Kerzeli Irantzu Anzar Donjete Simnica Elin Aamdal Tormod Guren Trevor Clancy Artur Mezheyeuski Else Marit Inderberg Sara M Mangsbo Mascha Binder Eivind Hovig Gustav Gaudernack	CONCLUSION: Clinical responses were observed irrespective of established predictive biomarkers for checkpoint inhibitor efficacy, indicating an added benefit of the vaccine-induced T cells. The clinical and immunological read-out warrants further investigation of UV1 in combination with checkpoint inhibitors. Trial registration Clinicaltrials.gov identifier: NCT02275416. Registered October 27, 2014. https://clinicaltrials.gov/ct2/show/NCT02275416?term=uv1&draw=2&rank=6.	pmid:36089578 doi:10.1186/s12967-022-03624-z	Sun, 11 Sep 2022 06:00:00 -0400
15	pubmed:36089604	Dynamic regulation of HIF-1 signaling in the rhesus monkey heart after ischemic injury	Tao Wang Ying Xiao Jingyao Zhang Fujia Jing Guodan Zeng	CONCLUSIONS: This study demonstrated a dynamic, functional-specific regulation of HIF-1 target gene expression during the progression of MI. The fine-tuning of HIF-1 signaling in the ischemic heart may be relate to the alteration in myocardial copper homeostasis. These findings provide transcriptomic insights into the distinct roles of HIF-1 signaling in the heart after ischemic injury, which will help determine the beneficial cutoff point for HIF-1 targeted therapy in ischemic heart diseases.	pmid:36089604 doi:10.1186/s12872-022-02841-0	Sun, 11 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
16	pubmed:36089633	Changing trends in the development of AAV-based gene therapies: a meta-analysis of past and present therapies	Tamara Burdett Samir Nuseibeh	Gene therapy has seen a transformation from a proof-of-concept approach to a clinical reality over the past several decades, with adeno-associated virus (AAV)-mediated gene therapy emerging as the leading platform for in vivo gene transfer. A systematic review of AAV-based gene therapies in clinical development was conducted herein to determine why only a handful of AAV-based gene therapy products have achieved market approval. The indication to be treated, route of administration and vector...	pmid:36089633 doi:10.1038/s41434-022-00363-0	Sun, 11 Sep 2022 06:00:00 -0400
17	pubmed:36089643	Treatment Patterns of Real-World Patients with TRK Fusion Cancer Treated by US Community Oncologists	Andrew J Klink Abhishek Kavati Awa Gassama Tom Kozlek Ajeet Gajra Ruth Antoine	CONCLUSION AND RELEVANCE: Among patients with advanced/metastatic TRK fusion solid tumors, medical oncologists reported that approximately two-thirds initiated a TRKi during the study period. Treatment with a TRKi was longer in duration compared to non-TRKi treatment in 1L and 2L therapy. Additional research is needed to gain insight into the association between early TRKi therapy initiation and clinical outcomes in the real-world setting.	pmid:36089643 doi:10.1007/s11523-022-00909-7	Sun, 11 Sep 2022 06:00:00 -0400
18	pubmed:36089668	Development of Surface Chemical Strategies for Synthesizing Redox-Responsive Diatomite Nanoparticles as a Green Platform for On-Demand Intracellular Release of an Antisense Peptide Nucleic Acid Anticancer Agent	Monica Terracciano Flavia Fontana Andrea Patrizia Falanga Stefano D'Errico Giulia Torrieri Francesca Greco Chiara Tramontano Ilaria Rea Gennaro Piccialli Luca De Stefano Giorgia Oliviero Hélder A Santos Nicola Borbone	Redox-responsive silica drug delivery systems are synthesized by aeco-friendly diatomite source to achieve on-demand release of peptide nucleic acid (PNA) in tumor reducing microenvironment, aiming to inhibit the immune checkpoint programmed cell death 1 receptor/programmed cell death receptor ligand 1 (PD-1/PD-L1) in cancer cells. The nanoparticles (NPs) are coated with polyethylene glycol chains as gatekeepers to improve their physicochemical properties and control drug release through the...	pmid:36089668 doi:10.1002/smll.202204732	Sun, 11 Sep 2022 06:00:00 -0400
19	pubmed:36089795	Identification of virulence markers and phylogenetic groups association, and Antimicrobial Susceptibility of uropathogenic Escherichia coli isolates	Dahbia Yasmina Meziani Nicolas Barnich Anouar Boucheham Mohamed Larbi Rezgoune Kaddour Benlabed Michael Rodrigues Dalila Satta	CONCLUSION: Certain virulence factors have been shown to be potential targets for drug design and therapeutic pathways in order to deal with the antimicrobial resistance problem enhanced by antibiotic therapy.	pmid:36089795 doi:10.2174/1871526522666220908161529	Mon, 12 Sep 2022 06:00:00 -0400
20	pubmed:36089809	Putaminal Recombinant Glucocerebrosidase Delivery with Magnetic Resonance-Guided Focused Ultrasound in Parkinson's Disease: A Phase I Study	Ying Meng Christopher B Pople Yuexi Huang Ryan M Jones Julie Ottoy Maged Goubran Lais M Oliveira Benjamin Davidson Liam S P Lawrence Angus Z Lau Allison Bethune Pejman Maralani Agessandro Abrahao Clement Hamani Kullervo Hynynen Suneil K Kalia Nir Lipsman Lorraine V Kalia	CONCLUSIONS: Results from this study demonstrate the safety and feasibility of MRgFUS GCase delivery in PD and support further investigation of this approach. © 2022 The Authors. Movement Disorders published by Wiley Periodicals LLC on behalf of International Parkinson and Movement Disorder Society.	pmid:36089809 doi:10.1002/mds.29190	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
21	pubmed:36089839	Neural signatures of default mode network in major depression disorder after electroconvulsive therapy	Yuanyuan Li Xiaohui Yu Yingzi Ma Jing Su Yue Li Shunli Zhu Tongjian Bai Qiang Wei Benjamin Becker Zhiyong Ding Kai Wang Yanghua Tian Jiaojian Wang	Functional abnormalities of default mode network (DMN) have been well documented in major depressive disorder (MDD). However, the association of DMN functional reorganization with antidepressant treatment and gene expression is unclear. Moreover, whether the functional interactions of DMN could predict treatment efficacy is also unknown. Here, we investigated the link of treatment response with functional alterations of DMN and gene expression with a comparably large sample including 46...	pmid:36089839 doi:10.1093/cercor/bhac311	Mon, 12 Sep 2022 06:00:00 -0400
22	pubmed:36090028	Characterizing the molecular heterogeneity of clear cell renal cell carcinoma subgroups classified by miRNA expression profile	Tao Shen Yingdong Song Xiangting Wang Haiyang Wang	Clear cell renal cell carcinoma (ccRCC) is a heterogeneous disease that is associated with poor prognosis. Recent works have revealed the significant roles of miRNA in ccRCC initiation and progression. Comprehensive characterization of ccRCC based on the prognostic miRNAs would contribute to clinicians' early detection and targeted treatment. Here, we performed unsupervised clustering using TCGA-retrieved prognostic miRNAs expression profiles. Two ccRCC subtypes were identified after assessing...	pmid:36090028 pmc:PMC9459094 doi:10.3389/fmolb.2022.967934	Mon, 12 Sep 2022 06:00:00 -0400
23	pubmed:36090045	Machine learning-based screening of an epithelial-mesenchymal transition-related long non-coding RNA signature reveals lower-grade glioma prognosis and the tumor microenvironment and predicts antitumor therapy response	Nan Wang Xin Gao Hang Ji Shuai Ma Jiasheng Wu Jiawei Dong Fang Wang Hongtao Zhao Zhihui Liu Xiuwei Yan Bo Li Jianyang Du Jiheng Zhang Shaoshan Hu	Epithelial-mesenchymal transition (EMT) confers high invasive and migratory capacity to cancer cells, which limits the effectiveness of tumor therapy. Long non-coding RNAs (lncRNAs) can regulate the dynamic process of EMT at different levels through various complex regulatory networks. We aimed to comprehensively analyze and screen EMT-related lncRNAs to characterize lower-grade glioma (LGG) tumor biology and provide new ideas for current therapeutic approaches. We retrieved 1065 LGG samples...	pmid:36090045 pmc:PMC9459009 doi:10.3389/fmolb.2022.942966	Mon, 12 Sep 2022 06:00:00 -0400
24	pubmed:36090054	Classification of stomach adenocarcinoma based on fatty acid metabolism-related genes frofiling	Chunhua Liu Yongjun Tao Huajian Lin Xiqiang Lou Simin Wu Liping Chen	Background: Fatty acid metabolism (FAM)-related genes play a key role in the development of stomach adenocarcinoma (STAD). Although immunotherapy has led to a paradigm shift in STAD treatment, the overall response rate of immunotherapy for STAD is low due to heterogeneity of the tumor immune microenvironment (TIME). How FAM-related genes affect TIME in STAD remains unclear. Methods: The univariate Cox regression analysis was performed to screen prognostic FAM-related genes using transcriptomic...	pmid:36090054 pmc:PMC9461144 doi:10.3389/fmolb.2022.962435	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
25	pubmed:36090113	Genomic characteristics of clinical multidrug-resistant *Proteus* isolates from a tertiary care hospital in southwest China	Ying Li Qian Liu Yichuan Qiu Chengju Fang Yungang Zhou Junping She Huan Chen Xiaoyi Dai Luhua Zhang	Multidrug-resistant (MDR) Proteus, especially those strains producing extended-spectrum -lactamases (ESBL) and carbapenemases, represents a major public health concern. In the present work, we characterized 27 MDR Proteus clinical isolates, including 23 Proteus mirabilis, three Proteus terrae, and one Proteus faecis, by whole-genome analysis. Among the 27 isolates analyzed, SXT/R391 ICEs were detected in 14 strains, and the complete sequences of nine ICEs were obtained. These ICEs share a...	pmid:36090113 pmc:PMC9449695 doi:10.3389/fmicb.2022.977356	Mon, 12 Sep 2022 06:00:00 -0400
26	pubmed:36090130	Outcome of pediatric chronic myeloid leukemia with management focusing on the monitoring of BCR-ABL fusion gene transcript levels	Ibrahim Al-Ghemlas Saad Al-Daama Hawazin Aqueel Khawar Siddiqui Hassan El-Solh Hala Omer Loloah AlRajeh Amal Al-Seraihy Ali Alahmari Hawazen AlSaedi Awatif AlAnazi Mouhab Ayas	CONCLUSION: We report an excellent outcome with an overall survival (OS) of 100% at 5-year and disease-free survival (DFS) of 91.7% (8.0%). All our patients achieved MMR and only one patient had loss of MMR on follow-up. Eight patients (66.7%) achieved complete molecular response (CMR).	pmid:36090130 pmc:PMC9441250 doi:10.1016/j.ijpam.2022.04.001	Mon, 12 Sep 2022 06:00:00 -0400
27	pubmed:36090157	Risk factors for inhibitors in hemophilia A based on RNA-seq and DNA methylation	Wei Liu Cuicui Lyu Wentian Wang Feng Xue Lingling Chen Huiyuan Li Ying Chi Yueshen Ma Runhui Wu Yunhai Fang Lei Zhang Renchi Yang	CONCLUSIONS: There is an upregulation of genes involved in activation of the immune system in hemophilia A patients with inhibitors. The lncRNA and methylation modifications may play important roles in inhibitor production. These findings are potentially to reveal novel therapeutic targets for prevention and treatment of inhibitors.	pmid:36090157 pmc:PMC9445143 doi:10.1002/rth2.12794	Mon, 12 Sep 2022 06:00:00 -0400
28	pubmed:36090221	Transplantation of IGF-1-induced BMSC-derived NPCs promotes tissue repair and motor recovery in a rat spinal cord injury model	Putri Nur Hidayah Al-Zikri Tee Jong Huat Amir Ali Khan Azim Patar Mohammed Faruque Reza Fauziah Mohamad Idris Jafri Malin Abdullah Hasnan Jaafar	Bone marrow-derived mesenchymal stem cells (BMSCs) have therapeutic potential for spinal cord injury (SCI). We have shown that insulin-like growth factor 1 (IGF-1) enhances the cellular proliferation and survivability of BMSCs-derived neural progenitor cells (NPCs) by downregulating miR-22-3p. However, the functional application of BMSCs-derived NPCs has not been investigated fully. In this study, we demonstrate that knockdown of endogenous miR-22-3p in BMSCs-derived NPCs upregulates Akt1...	pmid:36090221 pmc:PMC9449758 doi:10.1016/j.heliyon.2022.e10384	Mon, 12 Sep 2022 06:00:00 -0400
29	pubmed:36090572	Transient pseudohypoaldosteronism in infancy mainly manifested as poor appetite and vomiting: Two case reports and review of the literature	Yueerlanmu Tuoheti Yucan Zheng Yan Lu Mei Li Yu Jin	CONCLUSION: Transient pseudohypoaldosteronism should be considered in children younger than 6 months, presenting with vomiting, poor appetite, unexplained hyponatremia, hyperkalemia, elevated aldosterone levels, and urethral malformation or urinary tract infection. Furthermore, attention should be paid to whether salt supplementation or anti-infection therapy is effective.	pmid:36090572 pmc:PMC9452901 doi:10.3389/fped.2022.895647	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
30	pubmed:36090579	Case report: A study on the *de novo KMT2D* variant of Kabuki syndrome with Goodpasture's syndrome by whole exome sequencing	Shuolin Li Jing Liu Yuan Yuan Aizhen Lu Fang Liu Li Sun Quanli Shen Libo Wang	Kabuki syndrome (KS) is a rare genetic disorder characterized by dysmorphic facial features, skeletal abnormalities, and intellectual disability. KMT2D and KDM6A were identified as the main causative genes. To our knowledge, there exist no cases of KS, which were reported with pneumorrhagia. In this study, a 10-month-old male was diagnosed to have KS with typical facial features, skeletal anomalies, and serious postnatal growth retardation. Whole exome sequencing of the trio family revealed the...	pmid:36090579 pmc:PMC9459111 doi:10.3389/fped.2022.933693	Mon, 12 Sep 2022 06:00:00 -0400
31	pubmed:36090639	A retrospective study for prognostic significance of type II diabetes mellitus and hemoglobin A1c levels in non-small cell lung cancer patients treated with pembrolizumab	Yinchen Shen Jiaqi Li Huiping Qiang Yuqiong Lei Qing Chang Runbo Zhong Giulia Maria Stella Francesco Gelsomino Yeon Wook Kim Afaf Abed Jialin Qian Tianqing Chu	CONCLUSIONS: Among patients treated with 1st-line immunotherapy, a higher HbA1c level (6.5%) indicated dismal OS, while history of DM, baseline blood glucose levels, and glucose changes during the treatment process were not significantly associated with any of the outcomes.	pmid:36090639 pmc:PMC9459606 doi:10.21037/tlcr-22-493	Mon, 12 Sep 2022 06:00:00 -0400
32	pubmed:36090746	Genome editing-mediated knock-in of therapeutic genes ameliorates the disease phenotype in a model of hemophilia	Jeong Hyeon Lee Hye-Kyung Oh Beom Seok Choi Ho Hyeon Lee Kyu Jun Lee Un Gi Kim Jina Lee Hyerim Lee Geon Seong Lee Se Jun Ahn Jeong Pil Han Seokjoong Kim Su Cheong Yeom Dong Woo Song	Recently, clinical trials of adeno-associated virus-mediated replacement therapy have suggested long-term therapeutic effects for several genetic diseases of the liver, including hemophilia. However, there remain concerns regarding decreased therapeutic effects when the liver is regenerated or when physiological proliferation occurs. Although genome editing using the clustered regularly interspaced short palindromic repeats/Cas9 system provides an opportunity to solve this problem, low knock-in...	pmid:36090746 pmc:PMC9403902 doi:10.1016/j.omtn.2022.08.002	Mon, 12 Sep 2022 06:00:00 -0400
33	pubmed:36090755	Development of versatile allele-specific siRNAs able to silence all the dominant dynamin 2 mutations	Swati Dudhal Lylia Mekzine Bernard Prudhon Karishma Soocheta Bruno Cadot Kamel Mamchaoui Delphine Trochet Marc Bitoun	Dominant centronuclear myopathy (CNM) is a rare form of congenital myopathy associated with a wide clinical spectrum, from severe neonatal to milder adult forms. There is no available treatment for this disease due to heterozygous mutations in the DNM2 gene encoding Dynamin 2 (DNM2). Dominant DNM2 mutations also cause rare forms of Charcot-Marie-Tooth disease and hereditary spastic paraplegia, and deleterious DNM2 overexpression was noticed in several diseases. The proof of concept for therapy...	pmid:36090755 pmc:PMC9439966 doi:10.1016/j.omtn.2022.08.016	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
34	pubmed:36090760	Chemically modified *in-vitro*-transcribed mRNA encoding thrombopoietin stimulates thrombopoiesis in mice	Yu Zhang Xiaodong Xi Hang Yu Liuyan Yang Jinzhong Lin Wen Yang Junling Liu Xuemei Fan Yingjie Xu	The use of messenger RNA (mRNA) enables the transient production of therapeutic proteins with stable and predictable translational kinetics and without the risk of insertional mutagenesis. Recent findings highlight the enormous potential of mRNA-based therapeutics. Here, we describe the synthesis of chemically modified thrombopoietin (TPO) mRNA through in vitro transcription and in vivo delivery via lipid nanoparticles (LNPs). After delivery of TPO mRNA in mice, compared with normal...	pmid:36090760 pmc:PMC9440273 doi:10.1016/j.omtn.2022.08.017	Mon, 12 Sep 2022 06:00:00 -0400
35	pubmed:36090803	Targeted therapy for pediatric diffuse intrinsic pontine glioma: a single-center experience	Giada Del Baldo Andrea Carai Rachid Abbas Antonella Cacchione Mara Vinci Valentina Di Ruscio Giovanna Stefania Colafati Sabrina Rossi Francesca Diomedi Camassei Nicola Maestro Sara Temelso Giulia Pericoli Emmanuel De Billy Isabella Giovannoni Alessia Carboni Martina Rinelli Emanuele Agolini Alan Mackay Chris Jones Silvia Chiesa Mario Balducci Franco Locatelli Angela Mastronuzzi	CONCLUSION: Despite the small simple size of our study, our data suggest a prognostic advantage and a safe profile of targeted therapies in DIPG patients, and we strongly advocate to reconsider the role of biopsy for these patients.	pmid:36090803 pmc:PMC9459464 doi:10.1177/17588359221113693	Mon, 12 Sep 2022 06:00:00 -0400
36	pubmed:36090896	Systematic Pan-Cancer Analysis Identifies CDK1 as an Immunological and Prognostic Biomarker	Yaqi Yang Qin Liu Xiyuan Guo Qing Yuan Siji Nian Pengyuan Kang Zixi Xu Lin Li Yingchun Ye	Cyclin-dependent kinase 1 (CDK1) plays an important role in cancer development, progression, and the overall process of tumorigenesis. However, no pan-cancer analysis has been reported for CDK1, and the predictive role of CDK1 in immune checkpoint inhibitors (ICIs) therapy response remains unexplored. Thus, in this study, we first investigated the potential oncogenic role of CDK1 in 33 tumors by multidimensional bioinformatics analysis based on The Cancer Genome Atlas (TCGA) and Gene Expression...	pmid:36090896 pmc:PMC9452984 doi:10.1155/2022/8115474	Mon, 12 Sep 2022 06:00:00 -0400
37	pubmed:36090967	Robust pyroptosis risk score guides the treatment options and predicts the prognosis of bladder carcinoma	Dingshan Deng Fenglian Liu Zhi Liu Zuowei Wu Yunbo He ChunYu Zhang Xiongbin Zu Zhenyu Ou Yongjie Wang	CONCLUSIONS: This study developed and validated a robust pyroptosis risk score that can predict the clinical outcomes and TME immune phenotypes of BLCA. In summary, the pyroptosis risk score helps drive precision therapy in patients with BLCA.	pmid:36090967 pmc:PMC9450692 doi:10.3389/fimmu.2022.965469	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
38	pubmed:36090979	Natural history of type 1 diabetes on an immunodysregulatory background with genetic alteration in B-cell activating factor receptor: A case report	Biagio Di Lorenzo Lucia Pacillo Giulia Milardi Tatiana Jofra Silvia Di Cesare Jolanda Gerosa Ilaria Marzinotto Ettore Zapparoli Beatrice Rivalta Cristina Cifaldi Federica Barzaghi Carmela Giancotta Paola Zangari Novella Rapini Annalisa Deodati Giada Amodio Laura Passerini Paola Carrera Silvia Gregori Paolo Palma Andrea Finocchi Vito Lampasona Maria Pia Cicalese Riccardo Schiaffini Gigliola Di Matteo Ivan Merelli Matteo Barcella Alessandro Aiuti Lorenzo Piemonti Caterina Cancrini Georgia Fousteri	The immunological events leading to type 1 diabetes (T1D) are complex and heterogeneous, underscoring the necessity to study rare cases to improve our understanding. Here, we report the case of a 16-year-old patient who showed glycosuria during a regular checkup. Upon further evaluation, stage 2 T1D, autoimmune thrombocytopenic purpura (AITP), and common variable immunodeficiency (CVID) were diagnosed. The patient underwent low carb diet, losing > 8 kg, and was placed on Ig replacement therapy....	pmid:36090979 pmc:PMC9459137 doi:10.3389/fimmu.2022.952715	Mon, 12 Sep 2022 06:00:00 -0400
39	pubmed:36091017	Plasma exosomal IRAK1 can be a potential biomarker for predicting the treatment response to renin-angiotensin system inhibitors in patients with IgA nephropathy	Jianping Wu Xiaona Wei Jiajia Li Yangang Gan Rui Zhang Qianqian Han Peifen Liang Yuchun Zeng Qiongqiong Yang	CONCLUSIONS: Plasma exosomal IRAK1 can be a potential biomarker for predicting the treatment response of RASi in patients with IgAN.	pmid:36091017 pmc:PMC9459338 doi:10.3389/fimmu.2022.978315	Mon, 12 Sep 2022 06:00:00 -0400
40	pubmed:36091025	Innate immunity and immunotherapy for hemorrhagic shock	Qingxia Huang Song Gao Yao Yao Yisa Wang Jing Li Jinjin Chen Chen Guo Daqing Zhao Xiangyan Li	Hemorrhagic shock (HS) is a shock result of hypovolemic injury, in which the innate immune response plays a central role in the pathophysiology ofthe severe complications and organ injury in surviving patients. During the development of HS, innate immunity acts as the first line of defense, mediating a rapid response to pathogens or danger signals through pattern recognition receptors. The early and exaggerated activation of innate immunity, which is widespread in patients with HS, results in...	pmid:36091025 pmc:PMC9453212 doi:10.3389/fimmu.2022.918380	Mon, 12 Sep 2022 06:00:00 -0400
41	pubmed:36091034	Anti-factor H antibody and its role in atypical hemolytic uremic syndrome	Rupesh Raina Guneive Mangat Gordon Hong Raghav Shah Nikhil Nair Brian Abboud Sumedha Bagga Sidharth Kumar Sethi	Atypical hemolytic uremic syndrome (aHUS) an important form of a thrombotic microangiopathy (TMA) that can frequently lead to acute kidney injury (AKI). An important subset of aHUS is the anti-factor H associated aHUS. This variant of aHUS can occur due to deletion of the complement factor H genes, CFHR1 and CFHR3, along with the presence of anti-factor H antibodies. However, it is a point of interest to note that not all patients with anti-factor H associated aHUS have a CFHR1/R3 deletion....	pmid:36091034 pmc:PMC9448717 doi:10.3389/fimmu.2022.931210	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
42	pubmed:36091069	Precise somatic genome editing for treatment of inborn errors of immunity	Qingzhou Meng Haixiang Sun Jianghuai Liu	Rapid advances in high throughput sequencing have substantially expedited the identification and diagnosis of inborn errors of immunity (IEI). Correction of faulty genes in the hematopoietic stem cells can potentially provide cures for the majority of these monogenic immune disorders. Given the clinical efficacies of vector-based gene therapies already established for certain groups of IEI, the recently emerged genome editing technologies promise to bring safer and more versatile treatment...	pmid:36091069 pmc:PMC9459235 doi:10.3389/fimmu.2022.960348	Mon, 12 Sep 2022 06:00:00 -0400
43	pubmed:36091112	Bile is a reliable and valuable source to study cfDNA in biliary tract cancers	Zhanghui Li Yelei Liu Junhui Fu Joseph Mugaanyi Junrong Yan Caide Lu Jing Huang	CONCLUSION: The consistency of all genomic alterations and tumor tissue-determined genomic alteration in the bile supernatant/pellet was significantly higher than in plasma. Bile supernatants/pellets are better for genetic sequencing and may also have potential clinical value to guide targeted therapy and evaluate prognosis. Bile cfDNA may be a feasible substitute for tumor tissue in the genetic testing of patients with BTC.	pmid:36091112 pmc:PMC9459008 doi:10.3389/fonc.2022.961939	Mon, 12 Sep 2022 06:00:00 -0400
44	pubmed:36091118	The molecular mechanisms of ferroptosis and its role in glioma progression and treatment	Mengyang Lu Yuanshuai Zhou Linjuan Sun Shaheryar Shafi Nafees Ahmad Minxuan Sun Jun Dong	Ferroptosis is one of the programmed modes of cell death that has attracted widespread attention recently and is capable of influencing the developmental course and prognosis of many tumors. Glioma is one of the most common primary tumors of the central nervous system, but effective treatment options are very limited. Ferroptosis plays a critical role in the glioma progression, affecting tumor cell proliferation, angiogenesis, tumor necrosis, and shaping the immune-resistant tumor...	pmid:36091118 pmc:PMC9450584 doi:10.3389/fonc.2022.917537	Mon, 12 Sep 2022 06:00:00 -0400
45	pubmed:36091137	Circular RNAs regulate parental gene expression: A new direction for molecular oncology research	Haicun Wang Xin Gao Shaobo Yu Weina Wang Guanglin Liu Xingming Jiang Dongsheng Sun	CircRNAs have been the focus of research in recent years. They are differentially expressed in various human tumors and can regulate oncogenes and tumor suppressor genes expression through various mechanisms. The diversity, stability, evolutionary conservatism and cell- or tissue-specific expression patterns of circRNAs also endow them with important regulatory roles in promoting or inhibiting tumor cells malignant biological behaviors progression. More interestingly, emerging studies also found...	pmid:36091137 pmc:PMC9453195 doi:10.3389/fonc.2022.947775	Mon, 12 Sep 2022 06:00:00 -0400
46	pubmed:36091143	The SREBP-dependent regulation of cyclin D1 coordinates cell proliferation and lipid synthesis	Arwa Aldaalis Maria T Bengoechea-Alonso Johan Ericsson	The sterol regulatory-element binding protein (SREBP) family of transcription factors regulates cholesterol, fatty acid, and triglyceride synthesis and metabolism. However, they are also targeted by the ubiquitin ligase Fbw7, a major tumor suppressor, suggesting that they could regulate cell growth. Indeed, enhanced lipid synthesis is a hallmark of many human tumors. Thus, the SREBP pathway has recently emerged as a potential target for cancer therapy. We have previously demonstrated that one of...	pmid:36091143 pmc:PMC9451027 doi:10.3389/fonc.2022.942386	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
47	pubmed:36091162	DNA methylation regulators-related molecular patterns and tumor immune landscape in hepatocellular carcinoma	Dingli Song Zhenyu Zhou Jie Wu Tao Wei Guang Zhao Hong Ren Boxiang Zhang	Increasing evidence showed that the dysregulation of DNA methylation regulators is a decisive feature of almost all cancer types and affects tumor progressions. However, few studies focused on the underlying influences of DNA methylation regulators-related genes (DMRegs) in immune cell-infiltration characteristics, tumor microenvironment (TME) and immunotherapy in HCC patients. In our study, the alterations of DNA methylation regulators modification patterns (DMRPs) were clustered from...	pmid:36091162 pmc:PMC9459088 doi:10.3389/fonc.2022.877817	Mon, 12 Sep 2022 06:00:00 -0400
48	pubmed:36091401	The role of exercise-induced myokines in promoting angiogenesis	Chao Qi Xianjing Song He Wang Youyou Yan Bin Liu	Ischemic diseases are a major cause of mortality or disability in the clinic. Surgical or medical treatment often has poor effect on patients with tissue and organ ischemia caused by diffuse stenoses. Promoting angiogenesis is undoubtedly an effective method to improve perfusion in ischemic tissues and organs. Although many animal or clinical studies tried to use stem cell transplantation, gene therapy, or cytokines to promote angiogenesis, these methods could not be widely applied in the clinic...	pmid:36091401 pmc:PMC9459110 doi:10.3389/fphys.2022.981577	Mon, 12 Sep 2022 06:00:00 -0400
49	pubmed:36091591	Validation of the Anticolitis Efficacy of the Jian-Wei-Yu-Yang Formula	Jing Yan Yan Tang Wei Yu Lu Jiang Chen Liu Qi Li Zhiqiang Zhang Changlei Shao Yang Zheng Xihao Liu Xincheng Liu	CONCLUSION: The JW capsule attenuated the progression of murine colitis by a prompt resolution of inflammation and bloody stool and by re-establishing a microbiome profile that favors re-epithelization and prevents carcinogenesis.	pmid:36091591 pmc:PMC9451982 doi:10.1155/2022/9110704	Mon, 12 Sep 2022 06:00:00 -0400
50	pubmed:36091596	Integrated Microarray Analysis to Identify Genes and Small-Molecule Drugs Associated with Stroke Progression	Shasha Cui Yunfeng Zhao Menghui Huang Huan Zhang Wei Zhao Zhenhua Chen	Several blood biomarkers are now considered increasingly important for stratifying risk, monitoring disease progression, and evaluating the response to therapy in ischemic stroke. The purpose of the present study was to identify the key genes associated with ischemic stroke progression and elucidate the potential therapeutic small molecules. Microarray datasets related to stroke for GSE58294, GSE22255, and GSE16561 were obtained from the Gene Expression Omnibus (GEO) database. Differentially...	pmid:36091596 pmc:PMC9458405 doi:10.1155/2022/7634509	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
51	pubmed:36091706	*COL17A1* editing *via* homology-directed repair in junctional epidermolysis bullosa	Igor Petkovi Johannes Bischof Thomas Kocher Oliver Patrick March Bernadette Liemberger Stefan Hainzl Dirk Strunk Anna Maria Raninger Heide-Marie Binder Julia Reichelt Christina Guttmann-Gruber Verena Wally Josefina Piñón Hofbauer Johann Wolfgang Bauer Ulrich Koller	CONCLUSION: Here we present a gene editing approach capable of reducing end joining-generated repair products while increasing the level of seamless HDR-mediated gene repair outcomes, thereby providing a promising CRISPR/Cas9-based gene editing approach for JEB.	pmid:36091706 pmc:PMC9454317 doi:10.3389/fmed.2022.976604	Mon, 12 Sep 2022 06:00:00 -0400
52	pubmed:36091772	Insights on prospects of nano-siRNA based approaches in treatment of Cancer	Rajat Goyal Hitesh Chopra Inderbir Singh Kamal Dua Rupesh K Gautam	siRNA interference, commonly referred to as gene silence, is a biological mechanism that inhibits gene expression in disorders such as cancer. It may enhance the precision, efficacy, and stability of medicines, especially genetic therapies to some extent. However, obstacles such as the delivery of oligonucleotide drugs to inaccessible areas of the body and the prevalence of severe side effects must be overcome. To maximize their potential, it is thus essential to optimize their distribution to...	pmid:36091772 pmc:PMC9452808 doi:10.3389/fphar.2022.985670	Mon, 12 Sep 2022 06:00:00 -0400
53	pubmed:36091786	Emerging role of different DNA methyltransferases in the pathogenesis of cancer	Pengcheng Liu Fan Yang Lizhi Zhang Ying Hu Bangjie Chen Jianpeng Wang Lei Su Mingyue Wu Wenjian Chen	DNA methylation is one of the most essential epigenetic mechanisms to regulate gene expression. DNA methyltransferases (DNMTs) play a vital role in DNA methylation in the genome. In mammals, DNMTs act with some elements to regulate the dynamic DNA methylation patterns of embryonic and adult cells. Conversely, the aberrant function of DNMTs is frequently the hallmark in judging cancer, including total hypomethylation and partial hypermethylation of tumor suppressor genes (TSGs), which improve the...	pmid:36091786 pmc:PMC9453300 doi:10.3389/fphar.2022.958146	Mon, 12 Sep 2022 06:00:00 -0400
54	pubmed:36091829	Recent advances of IDH1 mutant inhibitor in cancer therapy	Wangqi Tian Weitong Zhang Yifan Wang Ruyi Jin Yuwei Wang Hui Guo Yuping Tang Xiaojun Yao	Isocitrate dehydrogenase (IDH) is the key metabolic enzyme that catalyzes the conversion of isocitrate to -ketoglutarate (-KG). Two main types of IDH1 and IDH2 are present in humans. In recent years, mutations in IDH have been observed in several tumors, including glioma, acute myeloid leukemia, and chondrosarcoma. Among them, the frequency of IDH1 mutations is higher than IDH2. IDH1 mutations have been shown to increase the conversion of -KG to 2-hydroxyglutarate (2-HG). IDH1...	pmid:36091829 pmc:PMC9449373 doi:10.3389/fphar.2022.982424	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
55	pubmed:36091860	Oncology during the New Coronavirus Infection Pandemic	A D Kaprin N S Sergeeva P V Shegai B Ya Alekseev	The COVID-19 pandemic has served as a catalyst for a whole layer of scientific research, including in Russia, where, since 2020, international multicenter studies have been conducted on the impact of the coronavirus infection on the course of oncological diseases, as well as on the development and application of new clinical methods in oncology. In the years 2020-2022, new methods of nuclear medicine based on the targeted effect of ionizing radiation of radiopharmaceuticals began to be actively...	pmid:36091860 pmc:PMC9447986 doi:10.1134/S1019331622040141	Mon, 12 Sep 2022 06:00:00 -0400
56	pubmed:36091935	Identification and Characterization of Genes Related to the Prognosis of Hepatocellular Carcinoma Based on Single-Cell Sequencing	Wenbiao Chen Feng Zhang Huixuan Xu Xianliang Hou Donge Tang Yong Dai	The heterogeneity of hepatocellular carcinoma (HCC) highlights the importance of precision therapy. In recent years, single-cell RNA sequencing has been used to reveal the expression of genes at the single-cell level and comprehensively study cell heterogeneity. This study combined big data analytics and single-cell data mining to study the influence of genes on HCC prognosis. The cells and genes closely related to the HCC were screened through single-cell RNA sequencing (71,915 cells, including...	pmid:36091935 pmc:PMC9454301 doi:10.3389/pore.2022.1610199	Mon, 12 Sep 2022 06:00:00 -0400
57	pubmed:36092022	Successful case of olaparib treatment for castration-resistant prostate cancer with multiple DNA repair gene mutations: Use of comprehensive genome profiling for treatment-refractory cases	Yukiyoshi Hirayama Minoru Kato Kaoru Kimura Taiyo Otoshi Takeshi Yamasaki Junji Uchida	Herein, we report a case of a 59-year-old man with advanced castration-resistant prostate cancer with rectal invasion. Multimodal treatment, including drug therapy, surgery, and radiation therapy was sequentially performed; however, lymph node metastases repeatedly occurred. Tumor genomic profiling using FoundationOne CDx identified pathogenic alterations in three DNA repair genes, including BRCA2 frameshift mutation. Olaparib, a poly-ADP ribose polymerase inhibitor, showed marked response....	pmid:36092022 pmc:PMC9460157 doi:10.1016/j.eucr.2022.102210	Mon, 12 Sep 2022 06:00:00 -0400
58	pubmed:36092250	Generation of GLA-knockout human embryonic stem cell lines to model peripheral neuropathy in Fabry disease	Christine R Kaneski John A Hanover Ulrike H Schueler Hoffman	Fabry disease is an X-linked glycolipid storage disorder caused by mutations in the GLA gene which result in a deficiency in the lysosomal enzyme alpha galactosidase A (AGA). As a result, the glycolipid substrate Gb3 accumulates in critical tissues and organs producing a progressive debilitating disease. In Fabry disease up to 80% of patients experience life-long neuropathic pain that is difficult to treat and greatly affects their quality of life. The molecular mechanisms by which deficiency of...	pmid:36092250 pmc:PMC9449667 doi:10.1016/j.ymgmr.2022.100914	Mon, 12 Sep 2022 06:00:00 -0400
59	pubmed:36092311	Bioinformatic-based mechanism identification of *E2F1*-related ceRNA and *E2F1* immunoassays in hepatocellular carcinoma	Wenlei Dong Chao Zhan	CONCLUSIONS: This study provides a valuable direction for researching transcription factor E2F1, which may be conducive in identifying research targets for HCC-related molecular biological therapy and immunotherapy in future.	pmid:36092311 pmc:PMC9459178 doi:10.21037/jgo-22-674	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
60	pubmed:36092319	The role of tumor-infiltrating B cells in the tumor microenvironment of hepatocellular carcinoma and its prognostic value: a bioinformatics analysis	Jixue Zou Chubin Luo Haoyang Xin Tongchun Xue Xiaoying Xie Rongxin Chen Lan Zhang	CONCLUSIONS: Tumor-infiltrating B cells potentially exert a tumor-suppressive function in the microenvironment of HCC and the higher levels of B cell infiltration are associated with a favorable outcome of HCC. Targeted activation of B cells may improve the tumor immune-targeted therapy.	pmid:36092319 pmc:PMC9459216 doi:10.21037/jgo-22-717	Mon, 12 Sep 2022 06:00:00 -0400
61	pubmed:36092325	A novel senescence-associated LncRNA signature predicts the prognosis and tumor microenvironment of patients with colorectal cancer: a bioinformatics analysis	Enmin Huang Tao Ma Junyi Zhou Ning Ma Weisheng Yang Chuangxiong Liu Zehui Hou Shuang Chen Zhen Zong Bing Zeng Yingru Li Taicheng Zhou	CONCLUSIONS: SenALSig can better predict survival and risk in CRC patients, as well as help develop new anti-cancer treatment strategies for CRC.	pmid:36092325 pmc:PMC9459181 doi:10.21037/jgo-22-721	Mon, 12 Sep 2022 06:00:00 -0400
62	pubmed:36092326	Let food be thy medicine: the role of diet in colorectal cancer: a narrative review	Ying Zheng Lingnan Meng Hao Liu Lijuan Sun Yongzhan Nie Qiong Wu Daiming Fan Mengbin Li	CONCLUSIONS: People at high risk of CRC and those with CRC are recommended to eat a plant-based diet rich in fruits, vegetables, and whole grains with appropriate DF intake and to avoid high levels of processed meat, red meat, and highly refined grains.	pmid:36092326 pmc:PMC9459199 doi:10.21037/jgo-22-32	Mon, 12 Sep 2022 06:00:00 -0400
63	pubmed:36092348	Characterization of somatic mutations and pathway alterations during hepatocellular carcinoma vascular invasion using next-generation sequencing	Qi Li Qifan Zhang Jinzhang Chen Mengya Zang Xiaoyun Hu Rong Li Jinlin Hou Jie Zhou	CONCLUSIONS: Somatic mutations and pathway changes associated with vascular invasion in HCC were identified. The discovery of the molecular drivers of vascular invasion in HCC provides novel insights that can help guide further patient diagnosis and personalized therapy.	pmid:36092348 pmc:PMC9459210 doi:10.21037/jgo-22-556	Mon, 12 Sep 2022 06:00:00 -0400
64	pubmed:36092360	Therapeutic efficacy of rscAAVrh74.miniCMV.*LIPA* gene therapy in a mouse model of lysosomal acid lipase deficiency	Patricia Lam Anna Ashbrook Deborah A Zygmunt Cong Yan Hong Du Paul T Martin	Lysosomal acid lipase deficiency (LAL-D) presents as one of two rare autosomal recessive diseases: Wolman disease (WD), a severe disorder presenting in infancy characterized by absent or very low LAL activity, and cholesteryl ester storage disease (CESD), a less severe, later onset disease form. Recent clinical studies have shown efficacy of enzyme replacement therapy for both forms of LAL-D; however, no gene therapy approach has yet been developed for clinical use. Here, we show that...	pmid:36092360 pmc:PMC9403906 doi:10.1016/j.omtm.2022.08.001	Mon, 12 Sep 2022 06:00:00 -0400
65	pubmed:36092361	Immortalized human myoblast cell lines for the delivery of therapeutic proteins using encapsulated cell technology	Aurelien Lathuiliere Remi Vernet Emily Charrier Muriel Urwyler Olivier Von Rohr Marie-Claude Belkouch Valentin Saingier Thomas Bouvarel Davy Guillarme Adrien Engel Patrick Salmon Thomas Laumonier Julien Grogg Nicolas Mach	Despite many promising results obtained in previous preclinical studies, the clinical development of encapsulated cell technology (ECT) for the delivery of therapeutic proteins from macrocapsules is still limited, mainly due to the lack of an allogeneic cell line compatible with therapeutic application in humans. In our work, we generated an immortalized human myoblast cell line specifically tailored for macroencapsulation. In the present report, we characterized the immortalized myoblasts and...	pmid:36092361 pmc:PMC9418741 doi:10.1016/j.omtm.2022.07.017	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
66	pubmed:36092365	Preclinical safety and efficacy of lentiviral-mediated gene therapy for leukocyte adhesion deficiency type I	Cristina Mesa-Núñez Carlos Damián María Fernández-García Begoña Díez Gayatri Rao Jonathan D Schwartz Ken M Law Julián Sevilla Paula Río Rosa Yáñez Juan A Bueren Elena Almarza	Leukocyte adhesion deficiency type I (LAD-I) is a primary immunodeficiency caused by mutations in the ITGB2 gene, which encodes for the CD18 subunit of (2)-integrins. Deficient expression of (2)-integrins results in impaired neutrophil migration in response to bacterial and fungal infections. Using a lentiviral vector (LV) that mediates a preferential myeloid expression of human CD18 (Chim.hCD18-LV), we first demonstrated that gene therapy efficiently corrected the phenotype of mice with...	pmid:36092365 pmc:PMC9418989 doi:10.1016/j.omtm.2022.07.015	Mon, 12 Sep 2022 06:00:00 -0400
67	pubmed:36092366	Full-length ATP7B reconstituted through protein *trans*-splicing corrects Wilson disease in mice	Agnese Padula Raffaella Petruzzelli Sasha A Philbert Stephanie J Church Federica Esposito Severo Campione Marcello Monti Filomena Capolongo Claudia Perna Edoardo Nusco Hartmut H Schmidt Alberto Auricchio Garth J S Cooper Roman Polishchuk Pasquale Piccolo	Wilson disease (WD) is a genetic disorder of copper homeostasis, caused by deficiency of the copper transporter ATP7B. Gene therapy with recombinant adeno-associated vectors (AAV) holds promises for WD treatment. However, the full-length human ATP7B gene exceeds the limited AAV cargo capacity, hampering the applicability of AAV in this disease context. To overcome this limitation, we designed a dual AAV vector approach using split intein technology. Split inteins catalyze seamless ligation of...	pmid:36092366 pmc:PMC9436707 doi:10.1016/j.omtm.2022.08.004	Mon, 12 Sep 2022 06:00:00 -0400
68	pubmed:36092368	Immunogenicity assessment of AAV-based gene therapies: An IQ consortium industry white paper	Tong-Yuan Yang Manuela Braun Wibke Lembke Fraser McBlane John Kamerud Stephen DeWall Edit Tarcsa Xiaodong Fang Lena Hofer Uma Kavita Vijay V Upreti Swati Gupta LiNa Loo Alison J Johnson Rakesh Kantilal Chandode Kay-Gunnar Stubenrauch Maya Vinzing Cindy Q Xia Vibha Jawa	Immunogenicity has imposed a challenge to efficacy and safety evaluation of adeno-associated virus (AAV) vector-based gene therapies. Mild to severe adverse events observed in clinical development have been implicated with host immune responses against AAV gene therapies, resulting in comprehensive evaluation of immunogenicity during nonclinical and clinical studies mandated by health authorities. Immunogenicity of AAV gene therapies is complex due to the number of risk factors associated with...	pmid:36092368 pmc:PMC9418752 doi:10.1016/j.omtm.2022.07.018	Mon, 12 Sep 2022 06:00:00 -0400
69	pubmed:36092658	Nanocarriers: A novel strategy for the delivery of CRISPR/Cas systems	Faranak Hejabi Mohammad Sadegh Abbaszadeh Shirinsadat Taji Andrew O'Neill Fatemeh Farjadian Mohammad Doroudian	In recent decades, clustered regularly interspaced short palindromic repeat/CRISPR-associated protein (CRISPR/Cas) has become one of the most promising genome-editing tools for therapeutic purposes in biomedical and medical applications. Although the CRISPR/Cas system has truly revolutionized the era of genome editing, the safe and effective delivery of CRISPR/Cas systems represents a substantial challenge that must be tackled to enable the next generation of genetic therapies. In addition,...	pmid:36092658 pmc:PMC9450496 doi:10.3389/fchem.2022.957572	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
70	pubmed:36092717	A PARP1-related prognostic signature constructing and PARP-1 inhibitors screening for glioma	Hui Li Zhenhua Wang Yuanyuan Hou Jianxin Xi Zhenqiang He Han Lu Zhishan Du Sheng Zhong Qunying Yang	The current standard treatments of glioma include surgical resection, supplemented with radiotherapy and chemotherapy, but the prognosis is poor. PARP-1 (Poly ADP-ribose polymerase 1) is a hot spot for cancer-targeted therapy and was reported to be significantly elevated in glioma. In this study, we analyzed the role of PARP-1 in DNA damage repair, constructed a PARP1-related DNA-repair prognostic signature (DPS), and screened targeted drugs for glioma. RNA-seq data of 639 glioma samples were...	pmid:36092717 pmc:PMC9450093 doi:10.3389/fcell.2022.916415	Mon, 12 Sep 2022 06:00:00 -0400
71	pubmed:36092720	Comprehensive analysis of GINS subunits prognostic value and ceRNA network in sarcoma	Chuqiao Zhou Zhuoyuan Chen Bo Xiao Cheng Xiang Aoyu Li Ziyue Zhao Hui Li	Background: The GINS complex, composed of GINS1/2/3/4 subunits, is an essential structure of Cdc45-MCM-GINS (CMG) helicase and plays a vital role in establishing the DNA replication fork and chromosome replication. Meanwhile, GINS genes have been associated with the poor prognosis of various malignancies. However, the abnormal expression of GINS genes and their diagnostic and prognostic value in sarcomas (SARC) remain unclear. Methods: Oncomine, Gene Expression Profiling Interactive Analysis...	pmid:36092720 pmc:PMC9462653 doi:10.3389/fcell.2022.951363	Mon, 12 Sep 2022 06:00:00 -0400
72	pubmed:36092734	Radiation-induced FAP + fibroblasts are involved in keloid recurrence after radiotherapy	Yan Gao Xue Hou Yuyin Dai Ting Yang Kexin Chen	Background: Keloid scars (KSs), which are composed of abnormal hyperplastic scar tissue, form during skin wound healing due to excessive fibroblast activation and collagen secretion. Although surgical resection and radiation therapy are used to prevent recurrence, KS recurrence rates range from 15 to 23%, and the underlying mechanism is unclear. Methods: To elucidate the mechanism of keloid recurrence, we established a PDX model and the grafts remained for over 20 weeks after transplantation on...	pmid:36092734 pmc:PMC9449371 doi:10.3389/fcell.2022.957363	Mon, 12 Sep 2022 06:00:00 -0400
73	pubmed:36092820	Differentiation of beta-like cells from human induced pluripotent stem cell-derived pancreatic progenitor organoids	Sergio Pedraza-Arevalo Ana-Maria Cujba Mario Enrique Alvarez-Fallas Rocio Sancho	Human induced pluripotent stem cells (hiPSCs) and organoids are important for modeling human development and disease in vitro. In this study, we describe a protocol to differentiate hiPSC toward pancreatic progenitor (PP) organoids and beta-like cells. We detail the expansion and seeding of hiPSC, PP differentiation, organoid expansion, and the differentiation of PP into beta cells. Upon differentiation, organoids contained beta, delta, and alpha cells. For complete details on the use and...	pmid:36092820 pmc:PMC9449863 doi:10.1016/j.xpro.2022.101656	Mon, 12 Sep 2022 06:00:00 -0400
74	pubmed:36092870	Identification of lactate metabolism-related subtypes and development of a lactate-related prognostic indicator of lung adenocarcinoma	Xiaoyan Chang Tong Lu Ran Xu Chenghao Wang Jiaying Zhao Linyou Zhang	Background: Increasing evidence supports that lactate plays an important role in tumor proliferation, invasion and within the tumor microenvironment (TME). This is particularly relevant in lung adenocarcinoma (LUAD). Therefore, there is a current need to investigate lactate metabolism in LUAD patients and how lactate metabolism is affected by different therapies. Methods: Data from LUAD patients were collected from The Cancer Genome Atlas (TCGA) and patients were divided into two subtypes...	pmid:36092870 pmc:PMC9449370 doi:10.3389/fgene.2022.949310	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
75	pubmed:36092879	Identification of the pyroptosis-related prognostic gene signature and characterization of tumor microenvironment infiltration in triple-negative breast cancer	Ji Liu Jianli Ma Qingyuan Zhang	Background: Triple-negative breast cancer remains a highly malignant disease due to the lack of specific targeted therapy and immunotherapy. A growing body of evidence supports the role of pyroptosis in tumorigenesis and prognosis, but further exploration is needed to improve our understanding of the tumor microenvironment in patients with triple-negative breast cancer. Methods: Consensus clustering analysis was performed to construct pattern clusters. A correlation analysis was conducted...	pmid:36092879 pmc:PMC9453819 doi:10.3389/fgene.2022.929870	Mon, 12 Sep 2022 06:00:00 -0400
76	pubmed:36092890	Pancancer analysis of a potential gene mutation model in the prediction of immunotherapy outcomes	Lishan Yu Caifeng Gong	Background: Immune checkpoint blockade (ICB) represents a promising treatment for cancer, but predictive biomarkers are needed. We aimed to develop a cost-effective signature to predict immunotherapy benefits across cancers. Methods: We proposed a study framework to construct the signature. Specifically, we built a multivariate Cox proportional hazards regression model with LASSO using 80% of an ICB-treated cohort (n = 1661) from MSKCC. The desired signature named SIGP was the risk score of the...	pmid:36092890 pmc:PMC9459043 doi:10.3389/fgene.2022.917118	Mon, 12 Sep 2022 06:00:00 -0400
77	pubmed:36092891	Molecular characterization, clinical relevance and immune feature of m7G regulator genes across 33 cancer types	Zhanzhan Li Yanyan Li Lin Shen Liangfang Shen Na Li	Over 170 RNA modifications have been identified after transcriptions, involving in regulation of RNA splicing, processing, translation and decay. Growing evidence has unmasked the crucial role of N-methyladenosine (m6A) in cancer development and progression, while, as a relative newly found RNA modification, N-methylguanosine (m7G) is also certified to participate in tumorigenesis via different catalytic machinery from that of m6A. However, system analysis on m7G RNA modification-related...	pmid:36092891 pmc:PMC9453236 doi:10.3389/fgene.2022.981567	Mon, 12 Sep 2022 06:00:00 -0400
78	pubmed:36092895	Characterization of aging cancer-associated fibroblasts draws implications in prognosis and immunotherapy response in low-grade gliomas	Zijian Zhou Jinhong Wei Lijun Kuang Ke Zhang Yini Liu Zhongming He Luo Li Bin Lu	Background: Due to the highly variable prognosis of low-grade gliomas (LGGs), it is important to find robust biomarkers for predicting clinical outcomes. Aging cancer-associated fibroblasts (CAFs) within the senescent stroma of a tumor microenvironment (TME) have been recently reported to play a key role in tumor development. However, there are few studies focusing on this topic in gliomas. Methods and Results: Based on the transcriptome data from TCGA and CGGA databases, we identified aging...	pmid:36092895 pmc:PMC9449154 doi:10.3389/fgene.2022.897083	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
79	pubmed:36092897	m^{5}C Regulator-mediated methylation modification clusters contribute to the immune microenvironment regulation of multiple myeloma	Hefei Ren Chang Liu Hongkun Wu Zhenhua Wang Sai Chen Xiaomin Zhang Jigang Ren Huiying Qiu Lin Zhou	Background: Multiple myeloma (MM) is a hematological malignancy in which plasma cells proliferate abnormally. 5-methylcytosine (mC) methylation modification is the primary epigenetic modification and is involved in regulating the occurrence, development, invasion, and metastasis of various tumors; however, its immunological functions have not been systematically described in MM. Thus, this study aimed to clarify the significance of mC modifications and how the immune microenvironment is linked...	pmid:36092897 pmc:PMC9453209 doi:10.3389/fgene.2022.920164	Mon, 12 Sep 2022 06:00:00 -0400
80	pubmed:36092901	NcRNA-mediated upregulation of CAMK2N1 is associated with poor prognosis and tumor immune infiltration of gastric cancer	Kaipeng Peng Xiangqing Ren Qian Ren	Gastric cancer (GC) is still notorious for its poor prognosis and aggressive characteristics. Though great developments have been made in diagnosis and therapy for GC, the prognosis of patient is still perishing. In this study, differentially expressed genes (DEGs) in GC were first screened using three Gene Expression Omnibus (GEO) datasets (GSE13911, GSE29998, and GSE26899). Second, The Cancer Genome Atlas (TCGA) and Genotype-Tissue Expression (GTEx) data were used to validate expression of...	pmid:36092901 pmc:PMC9452964 doi:10.3389/fgene.2022.888672	Mon, 12 Sep 2022 06:00:00 -0400
81	pubmed:36092920	Repression of enhancer RNA PHLDA1 promotes tumorigenesis and progression of Ewing sarcoma *via* decreasing infiltrating T-lymphocytes: A bioinformatic analysis	Runzhi Huang Dan Huang Siqiao Wang Shuyuan Xian Yifan Liu Minghao Jin Xinkun Zhang Shaofeng Chen Xi Yue Wei Zhang Jianyu Lu Huizhen Liu Zongqiang Huang Hao Zhang Huabin Yin	Background: The molecular mechanisms of EWS-FLI-mediating target genes and downstream pathways may provide a new way in the targeted therapy of Ewing sarcoma. Meanwhile, enhancers transcript non-coding RNAs, known as enhancer RNAs (eRNAs), which may serve as potential diagnosis markers and therapeutic targets in Ewing sarcoma. Materials and methods: Differentially expressed genes (DEGs) were identified between 85 Ewing sarcoma samples downloaded from the Treehouse database and 3 normal bone...	pmid:36092920 pmc:PMC9453160 doi:10.3389/fgene.2022.952162	Mon, 12 Sep 2022 06:00:00 -0400
82	pubmed:36092935	Comprehensive analysis reveals a 5-gene signature and immune cell infiltration in Alzheimer's disease with qPCR validation	Fanmao Jin Yuemei Xi De Xie Qiang Wang	Over 50 million people around the world currently are suffering from Alzheimer's disease (AD) without any effective therapy. Neuroinflammation plays a pivotal role in AD, which leads us to probe the profile of immune cell infiltration in AD. Here, we analyzed a microarray dataset (GSE44770) containing 115 AD and 115 control samples to determine biomarkers and immune infiltration characteristics of AD by multiple bioinformatics methods. First, we identified 3,840 DEGs (1892 upregulated and 1948...	pmid:36092935 pmc:PMC9454400 doi:10.3389/fgene.2022.913535	Mon, 12 Sep 2022 06:00:00 -0400
83	pubmed:36092958	The Prognostic Role of Cuproptosis in Head and Neck Squamous Cell Carcinoma Patients: A Comprehensive Analysis	Qin Ding Xiaochuan Chen Wenquan Hong Lihua Wang Wei Liu Sunqin Cai Xin Chen Jun Lu Sufang Qiu	CONCLUSION: Our study identified and validated novel cuproptosis-related biomarkers for HNSCC prognosis and screening, which offer better insights into developing accurate, reliable, and novel cancer therapies in the era of precision medicine.	pmid:36092958 pmc:PMC9463014 doi:10.1155/2022/9996946	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
84	pubmed:36092960	Molecular Polymorphisms of Vascular Endothelial Growth Factor Gene and Bronchopulmonary Dysplasia in Very Low Birth Weight Infants	Laura Filonzi Serafina Perrone Maria Luisa Tataranno Cinzia Magnani Harold Dadomo Anthea Bottoni Marina Vaghi Francesco Nonnis Marzano	CONCLUSIONS: Two single nucleotide polymorphisms within VEGF and VEGFR1 genes are not associated with BPD. Further researches are needed to reveal gene polymorphisms involved in vascular development as contributors to the onset of BPD.	pmid:36092960 pmc:PMC9458363 doi:10.1155/2022/2793846	Mon, 12 Sep 2022 06:00:00 -0400
85	pubmed:36093044	Genetic association and Mendelian randomization for hypothyroidism highlight immune molecular mechanisms	Samuel Mathieu Mewen Briend Erik Abner Christian Couture Zhonglin Li Yohan Bossé Sébastien Thériault Tõnu Esko Benoit J Arsenault Patrick Mathieu	We carried out a genome-wide association analysis including 51,194 cases of hypothyroidism and 443,383 controls. In total, 139 risk loci were associated to hypothyroidism with genes involved in lymphocyte function. Candidate genes associated with hypothyroidism were identified by using molecular quantitative trait loci, colocalization, and enhancer-promoter chromatin looping. Mendelian randomization (MR) identified 42 blood expressed genes and circulating proteins as candidate causal molecules...	pmid:36093044 pmc:PMC9460554 doi:10.1016/j.isci.2022.104992	Mon, 12 Sep 2022 06:00:00 -0400
86	pubmed:36093051	Multiplexed protein profiling reveals spatial subcellular signaling networks	Shuangyi Cai Thomas Hu Mythreye Venkatesan Mayar Allam Frank Schneider Suresh S Ramalingam Shi-Yong Sun Ahmet F Coskun	Protein-protein interaction networks are altered in multi-gene dysregulations in many disorders. Image-based protein multiplexing sheds light on signaling pathways to dissect cell-to-cell heterogeneity, previously masked by the bulk assays. Herein, we present a rapid multiplexed immunofluorescence (RapMIF) method measuring up to 25-plex spatial protein maps from cultures and tissues at subcellular resolution, providing combinatorial 272 pairwise and 1,360 tri-protein signaling states across 33...	pmid:36093051 pmc:PMC9460555 doi:10.1016/j.isci.2022.104980	Mon, 12 Sep 2022 06:00:00 -0400
87	pubmed:36093115	The tumor microenvironment of hepatocellular carcinoma and its targeting strategy by CAR-T cell immunotherapy	Zhang Guizhen Ji Guanchang Liu Liwen Wang Huifen Ren Zhigang Sun Ranran Yu Zujiang	Hepatocellular carcinoma (HCC) is the major subtype of liver cancer, which ranks sixth in cancer incidence and third in mortality. Although great strides have been made in novel therapy for HCC, such as immunotherapy, the prognosis remains less than satisfactory. Increasing evidence demonstrates that the tumor immune microenvironment (TME) exerts a significant role in the evolution of HCC and has a non-negligible impact on the efficacy of HCC treatment. In the past two decades, the success in...	pmid:36093115 pmc:PMC9452721 doi:10.3389/fendo.2022.918869	Mon, 12 Sep 2022 06:00:00 -0400
88	pubmed:36093134	Phenotypic vs. genetic cascade screening for familial hypercholesterolemia: A case report	Anastasia V Blokhina Alexandra I Ershova Alexey N Meshkov Anna V Kiseleva Marina V Klimushina Anastasia A Zharikova Evgeniia A Sotnikova Vasily E Ramensky Oxana M Drapkina	One of the most common autosomal dominant disorders is familial hypercholesterolemia (FH), causing premature atherosclerotic cardiovascular diseases and a high risk of death due to lifelong exposure to elevated low-density lipoprotein cholesterol (LDL-C) levels. FH has a proven arsenal of treatments and the opportunity for genetic diagnosis. Despite this, FH remains largely underdiagnosed worldwide. Cascade screening is a cost-effective method for the identification of new patients with FH and...	pmid:36093134 pmc:PMC9453448 doi:10.3389/fcvm.2022.982607	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
89	pubmed:36093136	Missense mutation of *SERPINC1* (p.Ser426Leu) in a young patient presenting as refractory and recurrent venous thromboembolism: A case report	Haixu Yu Xiaoyan Gai Jianli Wang Jinman Zhuang Wanwan Guo Rui Qiao Hong Zhu Yongchang Sun	Genetic and acquired risk factors are extremely important mechanisms in the development of venous thromboembolism (VTE). Inherited antithrombin (AT) deficiency due to mutations in the SERPINC1 gene is a well-known risk factor for genetic thrombophilia. In this case, we reported a 28-year young abroad student who presented with refractory and recurrent VTE in-hospital. This patient presented with a 2-month history of right lower limb pain and 1 week of fever. The ultrasound showed deep venous...	pmid:36093136 pmc:PMC9448915 doi:10.3389/fcvm.2022.903785	Mon, 12 Sep 2022 06:00:00 -0400
90	pubmed:36093155	Case report: Mexiletine suppresses ventricular arrhythmias in Andersen-Tawil syndrome	Jing Yang Kun Li Tingting Lv Ying Xie Fang Liu Ping Zhang	It is arduous to determine clinical solutions for Andersen-Tawil syndrome (ATS) in patients intolerant of -blocker. Here, we present the case of a 7-year-old boy with periodic paralysis and dysmorphic features who experienced syncope four times during exercise. His ECG revealed enlarged U waves and QU-prolongation associated with ATS-specific U wave patterns, frequent PVCs, and non-sustained bidirectional or polymorphic ventricular tachycardia. The genetic test showed a de novo missense R218W...	pmid:36093155 pmc:PMC9453449 doi:10.3389/fcvm.2022.992185	Mon, 12 Sep 2022 06:00:00 -0400
91	pubmed:36093169	Glyoxylase-1 combats dicarbonyl stress and right ventricular dysfunction in rodent pulmonary arterial hypertension	Sasha Z Prisco Lynn Hartweck Jennifer L Keen Neal Vogel Felipe Kazmirczak Megan Eklund Anna R Hemnes Evan L Brittain Kurt W Prins	CONCLUSION: Excess protein glycation promotes RV dysfunction in preclinical PAH, potentially through suppression of FAO.	pmid:36093169 pmc:PMC9452736 doi:10.3389/fcvm.2022.940932	Mon, 12 Sep 2022 06:00:00 -0400
92	pubmed:36093431	Therapeutic arteriogenesis by factor-decorated fibrin matrices promotes wound healing in diabetic mice	Rosalinda D'Amico Camilla Malucelli Andrea Uccelli Andrea Grosso Nunzia Di Maggio Priscilla S Briquez Jeffrey A Hubbell Thomas Wolff Lorenz Gürke Edin Mujagic Roberto Gianni-Barrera Andrea Banfi	Chronic wounds in type-2 diabetic patients present areas of severe local skin ischemia despite mostly normal blood flow in deeper large arteries. Therefore, restoration of blood perfusion requires the opening of arterial connections from the deep vessels to the superficial skin layer, that is, arteriogenesis. Arteriogenesis is regulated differently from microvascular angiogenesis and is optimally stimulated by high doses of Vascular Endothelial Growth Factor-A (VEGF) together with...	pmid:36093431 pmc:PMC9452813 doi:10.1177/20417314221119615	Mon, 12 Sep 2022 06:00:00 -0400
93	pubmed:36093523	Bioinformatics analysis and identification of genes and pathways involved in patients with Wilms tumor	Yufeng Li Haizhou Tang Zhenwen Huang Huaxing Qin Qin Cen Fei Meng Liang Huang Lifang Lin Jian Pu Di Yang	CONCLUSIONS: Our study revealed robust gene signatures in Wilms tumor. Dysregulations of the signaling pathways were associated with the development and progression of the Wilms tumor, and 10 hub genes may play important roles in its diagnosis and therapy.	pmid:36093523 pmc:PMC9459508 doi:10.21037/tcr-22-1847	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
94	pubmed:36093687	TET-2 mutations predict poor outcomes and are associated with unfavorable clinical-biological features in PTCL, not otherwise specified and angioimmunoblastic T-cell lymphoma in Brazilian patients	Luís Alberto de Pádua Covas Lage Guilherme Carneiro Barreto Hebert Fabricio Culler Jéssica Billar Cavalcante Lucas Bassolli de Oliveira Alves Luciana Nardinelli Israel Bendit Maria Cláudia Nogueira Zerbini Vanderson Rocha Juliana Pereira	CONCLUSION: Mutations in RHOA, TET-2 and DNMT3A were frequent in Brazilian patients with nPTCL. TET-2 mutations were associated with lower ORR for CHOP-like chemotherapy, decreased PFS and unfavorable clinical-biological characteristics in non-ALCL (PTCL, NOS and AITL). Further studies using a larger cohort may validate our findings.	pmid:36093687 doi:10.3233/CBM-220013	Mon, 12 Sep 2022 06:00:00 -0400
95	pubmed:36094043	Treatment of metastatic uveal melanoma in 2022: improved treatment regimens and improved prognosis	David Reichstein Anderson Brock Caressa Lietman Meredith McKean	PURPOSE OF REVIEW: Until recently, metastatic uveal melanoma was associated with essentially uniform fatality within months. However, recent developments in screening, improved understanding of the genetic underpinnings of metastatic disease, and pivotal medication approvals have improved the disease's rate of fatality.	pmid:36094043 doi:10.1097/ICU.0000000000000905	Mon, 12 Sep 2022 06:00:00 -0400
96	pubmed:36094106	Preclinical Evaluation of Foamy Virus Vector-Mediated Gene Addition in Human Hematopoietic Stem/Progenitor Cells for Correction of LAD-1	Richard H Smith Hanan Bloomer Danielle Fink Keyvan Keyvanfar Md Nasimuzzaman Fátima Sancheznieto Roop Dutta Kacey Guenther Bui Luigi J Alvarado Thomas R Bauer Dennis Hickstein David Russell Punam Malik Johannes Van Der Loo Steven L Highfill Douglas B Kuhns Mehdi Pirooznia Andre Larochelle	Ex vivo gene therapy procedures targeting hematopoietic stem and progenitor cells (HSPCs) predominantly utilize lentivirus-based vectors for gene transfer. We provide the first pre-clinical evidence of the therapeutic utility of a foamy virus vector (FVV) for the genetic correction of human leukocyte adhesion deficiency type 1 (LAD-1), an inherited primary immunodeficiency resulting from mutation of the 2 integrin common chain, CD18. CD34+ HSPCs isolated from a severely-affected LAD-1 patient...	pmid:36094106 doi:10.1089/hum.2022.065	Mon, 12 Sep 2022 06:00:00 -0400
97	pubmed:36094193	Low Diversity and Instability of the Sinus Microbiota over Time in Adults with Cystic Fibrosis	Catherine R Armbruster Kelvin Li Megan R Kiedrowski Anna C Zemke Jeffrey A Melvin John Moore Samar Atteih Adam C Fitch Matthew DuPont Christopher D Manko Madison L Weaver Jordon R Gaston John F Alcorn Alison Morris Barbara A Methé Stella E Lee Jennifer M Bomberger	Chronic rhinosinusitis (CRS) is a common, yet underreported and understudied manifestation of upper respiratory disease in people with cystic fibrosis (CF). Recently developed standard of care guidelines for the management of CF CRS suggest treatment of upper airway disease may ameliorate lower airway disease. We sought to determine whether changes to sinus microbial community diversity and specific taxa known to cause CF lung disease are associated with increased respiratory disease and...	pmid:36094193 doi:10.1128/spectrum.01251-22	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
98	pubmed:36094204	Genomic and Molecular Identification of Genes Contributing to the Caspofungin Paradoxical Effect in *Aspergillus fumigatus*	Shu Zhao Adela Martin-Vicente Ana Cristina Colabardini Lilian Pereira Silva David C Rinker Jarrod R Fortwendel Gustavo Henrique Goldman John G Gibbons	Aspergillus fumigatus is a deadly opportunistic fungal pathogen responsible for ~100,000 annual deaths. Azoles are the first line antifungal agent used against A. fumigatus, but azole resistance has rapidly evolved making treatment challenging. Caspofungin is an important second-line therapy against invasive pulmonary aspergillosis, a severe A. fumigatus infection. Caspofungin functions by inhibiting -1,3-glucan synthesis, a primary and essential component of the fungal cell wall. A phenomenon...	pmid:36094204 doi:10.1128/spectrum.00519-22	Mon, 12 Sep 2022 06:00:00 -0400
99	pubmed:36094220	Naturally Acquired Kelch13 Mutations in Plasmodium falciparum Strains Modulate *In Vitro* Ring-Stage Artemisinin-Based Drug Tolerance and Parasite Survival in Response to Hyperoxia	Sandra Duffy Vicky M Avery	The ring-stage survival assay was utilized to assess the impact of physiological hyperoxic stress on dihydroartemisinin (DHA) tolerance for a panel of Plasmodium falciparum strains with and without Kelch13 mutations. Strains without naturally acquired Kelch13 mutations or the postulated genetic background associated with delayed parasite clearance time demonstrated reduced proliferation under hyperoxic conditions in the subsequent proliferation cycle. Dihydroartemisinin tolerance in three...	pmid:36094220 doi:10.1128/spectrum.01282-21	Mon, 12 Sep 2022 06:00:00 -0400
100	pubmed:36094344	Intestinal Engineered Probiotics as Living Therapeutics: Chassis Selection, Colonization Enhancement, Gene Circuit Design, and Biocontainment	Yan Huang Xiaojun Lin Siyang Yu Ruiyue Chen Weizhao Chen	Intestinal probiotics are often used for the in situ treatment of diseases, such as metabolic disorders, tumors, and chronic inflammatory infections. Recently, there has been an increased emphasis on intelligent, customized treatments with a focus on long-term efficacy; however, traditional probiotic therapy has not kept up with this trend. The use of synthetic biology to construct gut-engineered probiotics as live therapeutics is a promising avenue in the treatment of specific diseases, such as...	pmid:36094344 doi:10.1021/acssynbio.2c00314	Mon, 12 Sep 2022 06:00:00 -0400
101	pubmed:36094557	Comprehensive analysis of DTYMK in pan-cancer and verification in lung adenocarcinoma	Yue Zhang Hao Wang Ying Liu Jing Yang Xiaoxiao Zuo Meilian Dong Zhigang Zhang Yonggang Shi Xubin Deng Yaoyong Lv	Previous document have reported that the DTYMK gene were involved in the progression of cancers. However, its significance in the analysis of pan-cancer and specific molecular mechanism were still poorly understood. In the current study, we conducted a comprehensive study of the DTYMK gene associated with its clinical relevance across a broad spectrum of human tumors. In addition, association among DTYMK gene and tumor immunogenic features was also explored. Considering the results of pan-cancer...	pmid:36094557 doi:10.1042/BSR20221170	Mon, 12 Sep 2022 06:00:00 -0400
102	pubmed:36094607	NREP is a Diagnostic and Prognostic Biomarker, and Promotes Gastric Cancer Cell Proliferation and Angiogenesis	Qian Li Lei Fu Daoyuan Wu Jufeng Wang	Neuronal regeneration related protein (NREP), also known as P311, has been reported to participate in multiple biological processes. The detection of tumor biomarker favored a non-invasive early entry for cancer diagnosis and disease monitoring to prevent its worsening symptoms. This study is intended to investigate the clinical roles of NREP in gastric cancer (GC) and its effect on gastric cancer cell proliferation and angiogenesis. Our results demonstrated that NREP was typically upregulated...	pmid:36094607 doi:10.1007/s10528-022-10276-7	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
103	pubmed:36094711	The challenges in the identification of Escherichia coli from environmental samples and their genetic characterization	Adriana Osiska Ewa Korzeniewska Agnieszka Korzeniowska-Kowal Anna Wzorek Monika Harnisz Piotr Jachimowicz Martyna Buta-Hubeny Wiktor Zieliski	Escherichia coli bacteria are an essential indicator in evaluations of environmental pollution, which is why they must be correctly identified. This study aimed to determine the applicability of various methods for identifying E. coli strains in environmental samples. Bacterial strains preliminary selected on mFc and Chromocult media as E. coli were identified using MALDI Biotyper techniques, based on the presence of genes characteristic of E. coli (uidA, uspA, yaiO), as well as by 16S rRNA gene...	pmid:36094711 doi:10.1007/s11356-022-22870-8	Mon, 12 Sep 2022 06:00:00 -0400
104	pubmed:36094767	Bacteriophage as a potential therapy to control antibiotic-resistant Pseudomonas aeruginosa infection through topical application onto a full-thickness wound in a rat model	Nouran Rezk Abdallah S Abdelsattar Doaa Elzoghby Mona M Agwa Mohamed Abdelmoteleb Rania G Aly Mohamed S Fayez Kareem Essam Bishoy M Zaki Ayman El-Shibiny	CONCLUSION: The phage ZCPA1 exhibited high lytic activity against MDR P. aeruginosa planktonic and biofilms. In addition, phage ZCPA1 showed complete wound healing in the rat model. Hence, this research demonstrates the potential of phage therapy as a promising alternative in treating MDR P. aeruginosa.	pmid:36094767 doi:10.1186/s43141-022-00409-1	Mon, 12 Sep 2022 06:00:00 -0400
105	pubmed:36094773	Aseptic meningitis in Fabry disease due to a novel GLA variant: an expanded phenotype?	Paulo Ribeiro Nóbrega João Lucas Araújo Morais Alliane Milliane Ferreira Alisson Dantas de Medeiros Beatrice Araújo Duarte Deborah Moreira Rangel Fabrício Oliveira Lima Anderson Rodrigues Brandão de Paiva Luciana Paim-Marques Fernando Kok André Luiz Santos Pessoa Pedro Braga-Neto Fernanda Martins Maia Carvalho	CONCLUSION: We described aseptic meningitis in a family with a novel GLA variant. Meningitis might be a common phenomenon in FD and not a particularity of this variant. Understanding the mechanisms underlying meningitis and its association with cerebrovascular events may lead to a new paradigm of treatment for stroke in these patients. Further prospective studies with CSF collection in patients with FD and recurrent headache could help to elucidate this question.	pmid:36094773 doi:10.1007/s10072-022-06388-y	Mon, 12 Sep 2022 06:00:00 -0400
106	pubmed:36094800	Unprotonatable and ROS-Sensitive Nanocarrier for NIR Spatially Activated siRNA Therapy with Synergistic Drug Effect	Shaohui Deng Shiyin Wang Zecong Xiao Du Cheng	Although small interfering RNA (siRNA) therapy has achieved great progress, unwanted gene inhibition in normal tissues severely limits its extensive clinical applications due to uncontrolled siRNA biodistribution. Herein, a spatially controlled siRNA activation strategy is developed to achieve tumor-specific siRNA therapy without gene inhibition in the normal tissues. The quaternary ammonium moieties are conjugated to amphiphilic copolymers via reactive oxygen species (ROS)-sensitive thioketal...	pmid:36094800 doi:10.1002/smll.202203823	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
107	pubmed:36094813	Web-Based Mindfulness-Based Interventions for Well-being: Randomized Comparative Effectiveness Trial	Louisa G Sylvia Mitchell R Lunn Juno Obedin-Maliver Robert N McBurney W Benjamin Nowell Rachel L Nosheny Richard A Mularski Millie D Long Peter A Merkel Mark J Pletcher Roberta E Tovey Christopher Scalchunes Rebecca Sutphen Ann S Martin Elizabeth J Horn Megan O'Boyle Lisa Pitch Michael Seid Susan Redline Sophie Greenebaum Nevita George Noah J French Caylin M Faria Nicha Puvanich Dustin J Rabideau Caitlin A Selvaggi Chu Yu Stephen V Faraone Shilpa Venkatachalam Debbe McCall Sharon F Terry Thilo Deckersbach Andrew A Nierenberg	CONCLUSIONS: Standard MBCT improved well-being but was not superior to a brief mindfulness intervention. This finding suggests that shorter mindfulness programs could yield important benefits across the general population of individuals with various medical conditions. Younger people and participants who completed more intervention sessions reported greater improvements in well-being, an effect that was more pronounced for participants in the MBCT condition. This finding suggests that standard...	pmid:36094813 doi:10.2196/35620	Mon, 12 Sep 2022 06:00:00 -0400
108	pubmed:36094837	Current updates on generations, approvals, and clinical trials of CAR T-cell therapy	Tadesse Asmamaw Dejenie Markeshaw Tiruneh G/Medhin Gashaw Dessie Terefe Fitalew Tadele Admasu Wondwossen Wale Tesega Endeshaw Chekol Abebe	Chimeric antigen receptor (CAR) T-cell therapy is a novel, customized immunotherapy that is considered a 'living' and self-replicating drug to treat cancer, sometimes resulting in a complete cure. CAR T-cells are manufactured through genetic engineering of T-cells by equipping them with CARs to detect and target antigen-expressing cancer cells. CAR is designed to have an ectodomain extracellularly, a transmembrane domain spanning the cell membrane, and an endodomain intracellularly. Since its...	pmid:36094837 doi:10.1080/21645515.2022.2114254	Mon, 12 Sep 2022 06:00:00 -0400
109	pubmed:36094907	Nanoparticle-mediated transgene expression of insulin-like growth factor 1 in the growth restricted guinea pig placenta increases placenta nutrient transporter expression and fetal glucose concentrations	Rebecca L Wilson Kristin Lampe Mukesh K Gupta Craig L Duvall Helen N Jones	Fetal growth restriction (FGR) significantly contributes to neonatal and perinatal morbidity and mortality. Currently, there are no effective treatment options for FGR during pregnancy. We have developed a nanoparticle gene therapy targeting the placenta to increase expression of human insulin-like growth factor 1 (hIGF1) to correct fetal growth trajectories. Using the maternal nutrient restriction guinea pig model of FGR, an ultrasound-guided, intraplacental injection of nonviral, polymer-based...	pmid:36094907 doi:10.1002/mrd.23644	Mon, 12 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
110	pubmed:36095221	ICBatlas: A comprehensive resource for depicting immune checkpoint blockade therapy characteristics from transcriptome profiles	Mei Yang Ya-Ru Miao Gui-Yan Xie Mei Luo Hui Hu Hang Fai Kwok Jian Feng An-Yuan Guo	Immune checkpoint blockade (ICB) therapy provides remarkable clinical benefits for multiple cancer types. Much work is currently being conducted to investigate the mechanisms of ICB therapy at the transcriptional level. Integrating the data produced by these studies will help us give more insight into the transcriptomic features of ICB therapy. We collected the transcriptome and clinical data of ICB-treated patient samples from the GEO, ArrayExpress, TCGA, and dbGaP databases. Based on the...	pmid:36095221 doi:10.1158/2326-6066.CIR-22-0249	Mon, 12 Sep 2022 06:00:00 -0400