gene therapy

	NCT Number	Title	Authors	Description	Identifier	Dates
1	pubmed:20301659	Pyridoxine-Dependent Epilepsy – *ALDH7A1*	Sidney M Gospe	CLINICAL CHARACTERISTICS: Pyridoxine-dependent epilepsy – ALDH7A1 (PDE-ALDH7A1) is characterized by seizures not well controlled with anti-seizure medication that are responsive clinically and electrographically to large daily supplements of pyridoxine (vitamin B(6)). This is true across a phenotypic spectrum that ranges from classic to atypical PDE-ALDH7A1. Intellectual disability is common, particularly in classic PDE-ALDH7A1.	pmid:20301659 nbk:NBK1486	Fri, 01 Jan 1993 06:00:00 -0500
2	pubmed:34978780	*SETD2* Neurodevelopmental Disorders	John Pappas Rachel Rabin	CLINICAL DESCRIPTION: SETD2 neurodevelopmental disorders (SETD2-NDDs) represent a clinical spectrum that most commonly includes various degrees of intellectual disability and behavioral problems (most typically an autism spectrum disorder), macrocephaly with or without ventriculomegaly, brain malformations (including Chiari 1 malformation and syringomyelia), and obesity with generalized overgrowth and advanced bone age. A specific, somewhat different phenotype (denoted SETD2-NDD with multiple...	pmid:34978780 nbk:NBK575927	Fri, 01 Jan 1993 06:00:00 -0500
3	pubmed:36126477	Ten-year follow-up of the observational RASTER study, prospective evaluation of the 70-gene signature in ER-positive, HER2-negative, node-negative, early breast cancer	Sonja B Vliek Florentine S Hilbers Agnes Jager Valesca P Retèl Jolien M Bueno de Mesquita Caroline A Drukker Sanne C Veltkamp Anneke M Zeillemaker Emiel J Rutgers Harm van Tinteren Wim H van Harten Laura J van 't Veer Marc J van de Vijver Sabine C Linn	CONCLUSIONS: These data confirm that clinically high-risk, genomically low-risk tumours have an excellent outcome in the real-world setting of shared decision-making. Together with the updated results of the MINDACT trial, these data support the use of the MammaPrint, in ER-positive, HER2-negative, node-negative, clinically high-risk breast cancer patients.	pmid:36126477 doi:10.1016/j.ejca.2022.07.036	Tue, 20 Sep 2022 06:00:00 -0400
4	pubmed:36126785	Spray drying siRNA-lipid nanoparticles for dry powder pulmonary delivery	Christoph M Zimmermann Domizia Baldassi Karen Chan Nathan B P Adams Alina Neumann Diana Leidy Porras-Gonzalez Xin Wei Nikolaus Kneidinger Mircea Gabriel Stoleriu Gerald Burgstaller Dominik Witzigmann Paola Luciani Olivia M Merkel	While all the siRNA drugs on the market target the liver, the lungs offer a variety of currently undruggable targets which could potentially be treated with RNA therapeutics. Hence, local, pulmonary delivery of RNA nanoparticles could finally enable delivery beyond the liver. The administration of RNA drugs via dry powder inhalers offers many advantages related to physical, chemical and microbial stability of RNA and nanosuspensions. The present study was therefore designed to test the...	pmid:36126785 doi:10.1016/j.jconrel.2022.09.021	Tue, 20 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
5	pubmed:36126853	Role of gut microbiota-derived branched-chain amino acids in the pathogenesis of Parkinson's disease: An animal study	Zhenzhen Yan Fan Yang Linlin Sun Jing Yu Lina Sun Yao Si Lifen Yao	Neuroinflammation caused by the disorder of gut microbiota and its metabolites is associated with the pathogenesis of Parkinson's disease (PD). Thus, it is necessary to identify certain molecules derived from gut microbiota to verify whether they could become intervention targets for the treatment of PD. The branched-chain amino acids (BCAAs), as a common dietary supplement, could modulate brain function. Herein, we investigated the longitudinal shifts of microbial community in mice treated with...	pmid:36126853 doi:10.1016/j.bbi.2022.09.009	Tue, 20 Sep 2022 06:00:00 -0400
6	pubmed:36126898	Novel epigenetic therapeutic strategies and targets in cancer	Quratulain Babar Ayesha Saeed Tanveer A Tabish Sabrina Pricl Helen Townley Nanasaheb Thorat	The critical role of dysregulated epigenetic pathways in cancer genesis, development, and therapy has typically been established as a result of scientific and technical innovations in next generation sequencing. RNA interference, histone modification, DNA methylation and chromatin remodelling are epigenetic processes that control gene expression without causing mutations in the DNA. Although epigenetic abnormalities are thought to be a symptom of cell tumorigenesis and malignant events that...	pmid:36126898 doi:10.1016/j.bbadis.2022.166552	Tue, 20 Sep 2022 06:00:00 -0400
7	pubmed:36126925	Congenital hemolytic anemias due to erythrocyte membrane and enzyme defects	Franziska Génevaux Annika Bertsch Lisa Wiederer Stefan Eber	Erythrocyte membrane and enzyme defects are the most common cause of congenital hemolytic anemias in the Central European population. Diagnostics include erythrocyte morphology, special biochemical tests such as osmotic fragility (AGLT) and EMA. For enzymopenic hemolytic anemias, cost-effective biochemical analysis remains the gold standard, supplemented by molecular genetic diagnostics when appropriate. Therapeutically, near complete splenectomy reduces hemolysis significantly for...	pmid:36126925 doi:10.1055/a-1767-8423	Tue, 20 Sep 2022 06:00:00 -0400
8	pubmed:36126995	Integrating specialist palliative care to improve care and reduce suffering: cystic fibrosis (InSPIRe:CF) - study protocol for a multicentre randomised clinical trial	Jane Lowers Elisabeth P Dellon Anne Stephenson Robert Arnold Andrew Althouse Kwonho Jeong Ethan Dubin Jesse Soodalter Cade Hovater Marie Bakitas Jessica Goggin William Hunt Sigrid Ladores Kimberly Curseen Gretchen Winter George Solomon Jonathan Ailon Douglas Conrad Dio Kavalieratos	INTRODUCTION: Cystic fibrosis (CF) is a life-limiting genetic disorder estimated to affect more than 160 000 individuals and their families worldwide. People living with CF commonly experience significant physical and emotional symptom burdens, disruptions to social roles and complex treatment decision making. While palliative care (PC) interventions have been shown to relieve many such burdens in other serious illnesses, no rigorous evidence exists for palliative care in CF. Thus, this study...	pmid:36126995 doi:10.1136/bmjresp-2022-001381	Tue, 20 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
9	pubmed:36127333	Single-cell transcriptome reveals cellular hierarchies and guides p-EMT-targeted trial in skull base chordoma	Qilin Zhang Lijiang Fei Rui Han Ruofan Huang Yongfei Wang Hong Chen Boyuan Yao Nidan Qiao Zhe Wang Zengyi Ma Zhao Ye Yichao Zhang Weiwei Wang Ye Wang Lin Kong Xuefei Shou Xiaoyun Cao Xiang Zhou Ming Shen Haixia Cheng Zhenwei Yao Chao Zhang Guoji Guo Yao Zhao	Skull base chordoma (SBC) is a bone cancer with a high recurrence rate, high radioresistance rate, and poorly understood mechanism. Here, we profiled the transcriptomes of 90,691 single cells, revealed the SBC cellular hierarchies, and explored novel treatment targets. We identified a cluster of stem-like SBC cells that tended to be distributed in the inferior part of the tumor. Combining radiated UM-Chor1 RNA-seq data and in vitro validation, we further found that this stem-like cell cluster is...	pmid:36127333 doi:10.1038/s41421-022-00459-2	Tue, 20 Sep 2022 06:00:00 -0400
10	pubmed:36127384	Development and verification of an immune-related gene prognostic index for gastric cancer	Chen Zhang Tao Liu Jian Wang JianTao Zhang	Immune checkpoint inhibitor (ICI) therapy is an emerging and effective approach to the treatment of gastric cancer (GC). However, the low response rate of GC patients to ICI therapy is a major limitation of ICI therapy. We investigated the transcriptomic signature of immune genes in GC could provide a comprehensive understanding of the tumor microenvironment (TME) and identify a valuable biomarker to predict the response of GC patients receiving immunotherapy. We performed the weighted gene...	pmid:36127384 doi:10.1038/s41598-022-20007-y	Tue, 20 Sep 2022 06:00:00 -0400
11	pubmed:36127509	Impact of conditioning chemotherapy on lymphocyte kinetics and outcomes in LBCL patients treated with CAR T-cell therapy	Paolo Strati Andrew P Jallouk Ryan Sun Jaihee Choi Kaberi Das Hua-Jay Cherng Sairah Ahmed Hun J Lee Swaminathan P Iyer Ranjit Nair Loretta J Nastoupil Raphael E Steiner Chad D Huff Yao Yu Haleigh Mistry Brittany Pulsifer Mansoor Noorani Neeraj Saini Elizabeth J Shpall Partow Kebriaei Christopher R Flowers Jason R Westin Michelle A T Hildebrandt Sattva S Neelapu	Conditioning chemotherapy (CCT) has been shown to be essential for optimal efficacy of chimeric antigen receptor (CAR) T-cell therapy. Here, we determined whether the change in absolute lymphocyte count, referred to as delta lymphocyte index (DLIx), may serve as a surrogate marker for pharmacodynamic effects of CCT and whether it associated with germline genetic variants in patients with large B-cell lymphoma (LBCL). One-hundred and seventy-one patients were included, of which 86 (50%) received...	pmid:36127509 doi:10.1038/s41375-022-01704-z	Tue, 20 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
12	pubmed:36127731	Expert consensus on the diagnosis and treatment of NTRK gene fusion solid tumors in China	Chunwei Xu Lu Si Wenxian Wang Ziming Li Zhengbo Song Qian Wang Aijun Liu Jinpu Yu Wenfeng Fang Wenzhao Zhong Zhijie Wang Yongchang Zhang Jingjing Liu Shirong Zhang Xiuyu Cai Anwen Liu Wen Li Ping Zhan Hongbing Liu Tangfeng Lv Liyun Miao Lingfeng Min Yu Chen Jingping Yuan Feng Wang Zhansheng Jiang Gen Lin Xingxiang Pu Rongbo Lin Weifeng Liu Chuangzhou Rao Dongqing Lv Zongyang Yu Lei Lei Xiaoyan Li Chuanhao Tang Chengzhi Zhou Junping Zhang Junli Xue Hui Guo Qian Chu Rui Meng Jingxun Wu Rui Zhang Xiao Hu Jin Zhou Zhengfei Zhu Yongheng Li Hong Qiu Fan Xia Yuanyuan Lu Xiaofeng Chen Rui Ge Enyong Dai Yu Han Weiwei Pan Jiancheng Luo Hongtao Jia Xiaowei Dong Fei Pang Kai Wang Liping Wang Youcai Zhu Yanru Xie Xinqin Lin Jing Cai Jia Wei Fen Lan Huijing Feng Lin Wang Yingying Du Wang Yao Xuefei Shi	Gene fusions can drive tumor development for multiple types of cancer. Currently, many drugs targeting gene fusions are being approved for clinical application. At present, tyrosine receptor kinase (TRK) inhibitors targeting neurotrophic tyrosine receptor kinase (NTRK) gene fusions are among the first "tumor agnostic" drugs approved for pan-cancer use. Representative TRK inhibitors, including larotrectinib and entrectinib, have shown high efficacy for many types of cancer. At the same time,...	pmid:36127731 doi:10.1111/1759-7714.14644	Tue, 20 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
13	pubmed:36127988	The Role of Gut-Microbiota in the Pathophysiology and Therapy of Irritable Bowel Syndrome: A Systematic Review	Bijay Shrestha Deepkumar Patel Hriday Shah Kerollos S Hanna Harkirat Kaur Mohammad S Alazzeh Abhay Thandavaram Aneeta Channar Ansh Purohit Sathish Venugopal	Irritable Bowel Syndrome (IBS) is one of the most prevalent chronic gastrointestinal diseases, which is characterized by recurrent abdominal pain and altered bowel habits. The pathophysiological mechanisms are not completely clear for IBS, multiple factors such as genetic, psychosocial, environmental, visceral hypersensitivity, low-grade inflammation, gastrointestinal motility changes, food components, and intestinal microbiota are thought to play a role in the disease process of IBS. The rapid...	pmid:36127988 pmc:PMC9477602 doi:10.7759/cureus.28064	Wed, 21 Sep 2022 06:00:00 -0400
14	pubmed:36128198	miR-378 associated with proliferation, migration and apoptosis properties in A549 cells and targeted NPNT in COPD	Guoqing Qian Qi Liao Guoxiang Li Fengying Yin	CONCLUSIONS: These findings suggest that miR-378 can regulate the proliferation, migration, and apoptosis of A549 cells and target NPNT. miR-378 increased in COPD lung tissues while NPNT decreased, and might prove a potential target for novel drug therapy.	pmid:36128198 pmc:PMC9482771 doi:10.7717/peerj.14062	Wed, 21 Sep 2022 06:00:00 -0400
15	pubmed:36128253	Hip Pain in Nonambulatory Children with Type-I or II Spinal Muscular Atrophy	Rewais B Hanna Nick Nahm Melissa A Bent Sarah Sund Karen Patterson Mary K Schroth Matthew A Halanski	CONCLUSIONS: This study is, to our knowledge, the largest investigation to date to assess hip pain among nonambulatory children with type-I or type-II SMA and suggests that symptoms rather than radiographs be utilized to direct care. These data will be crucial in assessing any effects that the new DMTs have on the natural history of hip pathology and pain in nonambulatory patients with SMA.	pmid:36128253 pmc:PMC9478277 doi:10.2106/JBJS.OA.22.00011	Wed, 21 Sep 2022 06:00:00 -0400
16	pubmed:36128418	Arrhythmogenic Right Ventricular Cardiomyopathy: The Role of Genetics in Diagnosis, Management, and Screening	Mihir Odak Steven Douedi Anton Mararenko Abbas Alshami Islam Elkherpitawy Hani Douedi Eran Zacks Brett Sealove	Arrhythmogenic right ventricular cardiomyopathy (ARVC) is a predominantly autosomal dominant genetic condition in which fibrous and fatty tissue infiltrate and replace healthy myocardial tissue. This uncommon yet debilitating condition can cause ventricular arrhythmias, cardiac failure, and sudden cardiac death. Management focuses primarily on prevention of syndrome sequelae in order to prevent morbidity and mortality. Genetic testing and screening in affected families, although utilized...	pmid:36128418 pmc:PMC9451588 doi:10.14740/cr1373	Wed, 21 Sep 2022 06:00:00 -0400
17	pubmed:36128444	Oncogenic role of TWF2 in human tumors: A pan-cancer analysis	Wenjie Liu Gengwei Huo Peng Chen	To develop effective medicines, researchers must first understand the common and distinct mechanisms that drive oncogenic processes in human cancers. TWF1 and TWF2 belong to the actin-depolymerizing factor homology family. TWF1 has been identified as an important gene in lung, breast, and pancreatic cancer in recent investigations. TWF2's role in cancer remains largely unknown, no comprehensive pan-cancer studies have been conducted. We utilized the The Cancer Genome Atlas and Gene Expression...	pmid:36128444 pmc:PMC9449685 doi:10.1515/med-2022-0547	Wed, 21 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
18	pubmed:36128554	Generic model for biological regulation	Mauno Vihinen	A substantial portion of molecules in an organism are involved in regulation of a wide spectrum of biological processes. Several models have been presented for various forms of biological regulation, including gene expression regulation and physiological regulation; however, a generic model is missing. Recently a new unifying theory in biology, poikilosis, was presented. Poikilosis indicates that all systems display intrinsic heterogeneity, which is a normal state. The concept of poikilosis...	pmid:36128554 pmc:PMC9468631 doi:10.12688/f1000research.110944.1	Wed, 21 Sep 2022 06:00:00 -0400
19	pubmed:36128656	Glucocorticoids mediate transcriptome-wide alternative polyadenylation: Potential mechanistic and clinical implications	Thanh Thanh L Nguyen Duan Liu Huanyao Gao Zhenqing Ye Jeong-Heon Lee Lixuan Wei Jia Yu Lingxin Zhang Liewei Wang Tamas Ordog Richard M Weinshilboum	Alternative polyadenylation (APA) is a common genetic regulatory mechanism that generates distinct 3' ends for RNA transcripts. Changes in APA have been associated with multiple biological processes and disease phenotypes. However, the role of hormones and their drug analogs in APA remains largely unknown. In this study, we investigated transcriptome-wide the impact of glucocorticoids on APA in 30 human B-lymphoblastoid cell lines. We found that glucocorticoids could regulate APA for a subset of...	pmid:36128656 doi:10.1111/cts.13402	Wed, 21 Sep 2022 06:00:00 -0400
20	pubmed:36128718	Maternal diabetes negatively impacts fetal health	Cecilia González Corona Ronald J Parchem	Diabetes is a chronic metabolic disease affecting an increasing number of people. Although diabetes has negative health outcomes for diagnosed individuals, a population at particular risk are pregnant women, as diabetes impacts not only a pregnant woman's health but that of her child. In this review, we cover the current knowledge and unanswered questions on diabetes affecting an expectant mother, focusing on maternal and fetal outcomes.	pmid:36128718 doi:10.1098/rsob.220135	Wed, 21 Sep 2022 06:00:00 -0400
21	pubmed:36128835	Novel approach of desensitization in allergic reaction to Olaparib	Björn M Beurer Luise M Sprenger Kristina Graneß Freia Feldmann Ulrich Warnke Maria G Biersack Dorothea Fischer	CONCLUSION: Desensitization in a two-day suspension protocol is a safe method that ensures effective maintenance therapy for patients with allergic reactions to PARP inhibitors.	pmid:36128835 doi:10.1177/10781552221124041	Wed, 21 Sep 2022 06:00:00 -0400
22	pubmed:36128851	Current Status and Prospects of Clinical Treatment of Osteosarcoma	Zong-Yuan Jiang Ji-Bin Liu Xiao-Feng Wang Yu-Shui Ma Da Fu	Osteosarcoma, one of the common malignant tumors in the skeletal system, originates in mesenchymal tissue, and the most susceptible area of occurrence is the metaphysis with its abundant blood supply. Tumors are characterized by highly malignant spindle stromal cells that can produce bone-like tissue. Most of the osteosarcoma are primary, and a few are secondary. Osteosarcoma occurs primarily in children and adolescents undergoing vigorous bone growth and development. Most cases involve rapid...	pmid:36128851 doi:10.1177/15330338221124696	Wed, 21 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
23	pubmed:36128923	Tailoring head-tail mesoporous silica nanoparticles for enhanced gene transfection	Weixi Wu Anh Ngo Wenhuang Ban Yuening Zhong Dan Cheng Zhengying Gu Chengzhong Yu Hao Song	Plasmid DNA (pDNA) delivery has attracted extensive research interest due to its great potential in gene therapy. The design of efficient nano-vectors to promote cellular delivery and transfection of gene molecules is the key to success. Compared to conventional nanocarriers with spherical geometry, asymmetric nanoparticles have been well documented showing enhanced cellular uptake and drug delivery capability. However, the impact of asymmetric nanostructures on pDNA binding and following...	pmid:36128923 doi:10.1039/d2tb01737g	Wed, 21 Sep 2022 06:00:00 -0400
24	pubmed:36128934	A comprehensive overview of CRISPR/Cas 9 technology and application thereof in drug discovery	Amit Khurana Nilofer Sayed Vishakha Singh Isha Khurana Prince Allawadhi Pushkar Singh Rawat Umashanker Navik Sravan Kumar Pasumarthi Kala Kumar Bharani Ralf Weiskirchen	Clustered Regularly Interspaced Short Palindromic Repeat (CRISPR)-Cas technology possesses revolutionary potential to positively affect various domains of drug discovery. It has initiated a rise in the area of genetic engineering and its advantages range from classical science to translational medicine. These genome editing systems have given a new dimension to our capabilities to alter, detect and annotate specified gene sequences. Moreover, the ease, robustness and adaptability of the...	pmid:36128934 doi:10.1002/jcb.30329	Wed, 21 Sep 2022 06:00:00 -0400
25	pubmed:36129048	An update on the molecular biology of glioblastoma, with clinical implications and progress in its treatment	Elena Verdugo Iker Puerto Miguel Ángel Medina	Glioblastoma multiforme (GBM) is the most aggressive and common malignant primary brain tumor. Patients with GBM often have poor prognoses, with a median survival of 15 months. Enhanced understanding of the molecular biology of central nervous system tumors has led to modifications in their classifications, the most recent of which classified these tumors into new categories and made some changes in their nomenclature and grading system. This review aims to give a panoramic view of the last 3...	pmid:36129048 doi:10.1002/cac2.12361	Wed, 21 Sep 2022 06:00:00 -0400
26	pubmed:36129137	[Corrigendum] TP53 and RET may serve as biomarkers of prognostic evaluation and targeted therapy in hepatocellular carcinoma	Song Ye Xin-Yi Zhao Xiao-Ge Hu Tang Li Qiu-Ran Xu Huan-Ming Yang Dong-Sheng Huang Liu Yang	Subsequently to the publication of the above article, the authors have realized that a couple of clerical errors were made when writing the article, and wish to correct these errors in a corrigendum statement. First, in the Materials and methods section on p. 2216, the final sentence of the 'Immunohistochemistry and tissue microarray' subsection, the authors wish to add a further definition, so that the text reads as follows (changes highlighted in bold): 'The positive expression of RET was...	pmid:36129137 doi:10.3892/or.2022.8411	Wed, 21 Sep 2022 06:00:00 -0400
27	pubmed:36129287	Characterization of Phage Resistance and Their Impacts on Bacterial Fitness in Pseudomonas aeruginosa	Na Li Yigang Zeng Mengran Wang Rong Bao Yu Chen Xiaoyu Li Jue Pan Tongyu Zhu Bijie Hu Demeng Tan	The emergence and spread of antibiotic resistance pose serious environmental and health challenges. Attention has been drawn to phage therapy as an alternative approach to combat antibiotic resistance with immense potential. However, one of the obstacles to phage therapy is phage resistance, and it can be acquired through genetic mutations, followed by consequences of phenotypic variations. Therefore, understanding the mechanisms underlying phage-host interactions will provide us with greater...	pmid:36129287 doi:10.1128/spectrum.02072-22	Wed, 21 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
28	pubmed:36129301	Microevolution of CG23-I Hypervirulent Klebsiella pneumoniae during Recurrent Infections in a Single Patient	Yao-Chen Wang Min-Chi Lu Yia-Ting Li Hui-Ling Tang Pei-Yi Hsiao Bo-Han Chen Ru-Hsiou Teng Chien-Shun Chiou Yi-Chyi Lai	CG23-I lineage constitutes the majority of hypervirulent Klebsiella pneumoniae. A diabetic patient suffered six episodes of infections caused by CG23-I K. pneumoniae. A total of nine isolates were collected in 2020. We performed whole-genome sequencing to elucidate the within-patient evolution of CG23-I K. pneumoniae. The maximum pairwise difference among the nine longitudinally collected isolates was five single nucleotide polymorphisms. One of the mutations was at the Asp87 position of GyrA....	pmid:36129301 doi:10.1128/spectrum.02077-22	Wed, 21 Sep 2022 06:00:00 -0400
29	pubmed:36129635	A Novel Immunotoxin Targeting Epithelial Cell Adhesion Molecule Using Single Domain Antibody Fused to Diphtheria Toxin	Reyhaneh Roshan Shamsi Naderi Mahdi Behdani Reza Ahangari Cohan Fatemeh Kazemi-Lomedasht	Epithelial Cell Adhesion Molecule (EpCAM) is overexpressed in a variety of cancers such as colon, stomach, pancreas, and prostate adenocarcinomas. Inhibition of EpCAM is considered as a potential target for cancer therapy. In current study, anti-EpCAM immunotoxin (-EpCAM IT) was developed using genetic fusion of -EpCAM single domain antibody (nanobody) (-EpCAM Nb) to truncated form of diphtheria toxin. The expression of recombinant -EpCAM IT was induced by Isopropyl...	pmid:36129635 doi:10.1007/s12033-022-00565-2	Wed, 21 Sep 2022 06:00:00 -0400
30	pubmed:36129672	Pyroptosis: a novel signature to predict prognosis and immunotherapy response in gliomas	Guiying He Zhimin Chen Shenghua Zhuo Jingzhi Tang Weijie Hao Kun Yang Chunshui Yang	Gliomas are the most common primary brain tumors and are highly malignant with a poor prognosis. Pyroptosis, an inflammatory form of programmed cell death, promotes the inflammatory cell death of cancer. Studies have demonstrated that pyroptosis can promote the inflammatory cell death (ICD) of cancer, thus affecting the prognosis of cancer patients. Therefore, genes that control pyroptosis could be a promising candidate bio-indicator in tumor therapy. The function of pyroptosis-related genes...	pmid:36129672 doi:10.1007/s13577-022-00791-5	Wed, 21 Sep 2022 06:00:00 -0400
31	pubmed:36129940	Computational gene expression analysis reveals distinct molecular subgroups of T-cell prolymphocytic leukemia	Nathan Mikhaylenko Linus Wahnschaffe Marco Herling Ingo Roeder Michael Seifert	T-cell prolymphocytic leukemia (T-PLL) is a rare blood cancer with poor prognosis. Overexpression of the proto-oncogene TCL1A and missense mutations of the tumor suppressor ATM are putative main drivers of T-PLL development, but so far only little is known about the existence of T-PLL gene expression subtypes. We performed an in-depth computational reanalysis of 68 gene expression profiles of one of the largest currently existing T-PLL patient cohorts. Hierarchical clustering combined with...	pmid:36129940 doi:10.1371/journal.pone.0274463	Wed, 21 Sep 2022 06:00:00 -0400
32	pubmed:36129942	CYCLIN K down-regulation induces androgen receptor gene intronic polyadenylation, variant expression and PARP inhibitor vulnerability in castration-resistant prostate cancer	Rui Sun Ting Wei Donglin Ding Jianong Zhang Sujun Chen Housheng Hansen He Liguo Wang Haojie Huang	Androgen receptor (AR) messenger RNA (mRNA) alternative splicing variants (AR-Vs) are implicated in castration-resistant progression of prostate cancer (PCa), although the molecular mechanism underlying the genesis of AR-Vs remains poorly understood. The CDK12 gene is often deleted or mutated in PCa and CDK12 deficiency is known to cause homologous recombination repair gene alteration or BRCAness via alternative polyadenylation (APA). Here, we demonstrate that pharmacological inhibition or...	pmid:36129942 doi:10.1073/pnas.2205509119	Wed, 21 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
33	pubmed:36129972	Semirational bioengineering of AAV vectors with increased potency and specificity for systemic gene therapy of muscle disorders	Jihad El Andari Edith Renaud-Gabardos Warut Tulalamba Jonas Weinmann Louise Mangin Quang Hong Pham Susanne Hille Antonette Bennett Esther Attebi Emanuele Bourges Christian Leborgne Nicolas Guerchet Julia Fakhiri Chiara Krämer Ellen Wiedtke Robert McKenna Laurence Guianvarc'h Magali Toueille Giuseppe Ronzitti Matthias Hebben Federico Mingozzi Thierry VandenDriessche Mavis Agbandje-McKenna Oliver J Müller Marinee K Chuah Ana Buj-Bello Dirk Grimm	Bioengineering of viral vectors for therapeutic gene delivery is a pivotal strategy to reduce doses, facilitate manufacturing, and improve efficacy and patient safety. Here, we engineered myotropic adeno-associated viral (AAV) vectors via a semirational, combinatorial approach that merges AAV capsid and peptide library screens. We first identified shuffled AAVs with increased specificity in the murine skeletal muscle, diaphragm, and heart, concurrent with liver detargeting. Next, we boosted...	pmid:36129972 doi:10.1126/sciadv.abn4704	Wed, 21 Sep 2022 06:00:00 -0400
34	pubmed:36130036	Plasmid-Based Donor Templates for Nonviral CRISPR/Cas9-Mediated Gene Knock-In in Human T Cells	Kate Senger Ilseyar Akhmetzyanova Benjamin Haley Sascha Rutz Soyoung A Oh	Effective and precise gene editing of T lymphocytes is critical for advancing the understanding of T cell biology and the development of next-generation cellular therapies. Although methods for effective CRISPR/Cas9-mediated gene knock-out in primary human T cells have been developed, complementary techniques for nonviral gene knock-in can be cumbersome and inefficient. Here, we report a simple and efficient method for nonviral CRISPR/Cas9-based gene knock-in utilizing plasmid-based donor DNA...	pmid:36130036 doi:10.1002/cpz1.538	Wed, 21 Sep 2022 06:00:00 -0400
35	pubmed:36130085	d-StructMAn: Containerized structural annotation on the scale from genetic variants to whole proteomes	Alexander Gress Sanjay K Srikakulam Sebastian Keller Vasily Ramensky Olga V Kalinina	CONCLUSIONS: d-StructMAn is the first of its kind and a highly efficient tool for structural annotation of protein-coding genetic variation in the context of observed and potential intermolecular interactions. d-StructMAn is readily applicable to proteome-scale datasets and can be an instrumental building machine-learning tool for predicting genotype-to-phenotype relationships.	pmid:36130085 doi:10.1093/gigascience/giac086	Wed, 21 Sep 2022 06:00:00 -0400
36	pubmed:36130114	Erratum to: Pharmacogenetics-guided dalcetrapib therapy after an acute coronary syndrome: the dal-GenE trial		No abstract	pmid:36130114 doi:10.1093/eurheartj/ehac534	Wed, 21 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
37	pubmed:36130152	Chimeric Antigen Receptor T-Cell Therapies: Barriers and Solutions to Access	Joseph Mikhael Jessica Fowler Nina Shah	Chimeric antigen receptor T-cell (CAR-T) therapies are relatively new treatments for patients with heavily pretreated hematologic malignancies. Although these innovative therapies can offer substantial benefit to patients with limited alternative treatment options, patient-access barriers exist. Conventional clinical trials are time-consuming and may be limited by strict patient eligibility criteria, resources, and availability of enrollment slots. Because of the complexity of the CAR-T...	pmid:36130152 doi:10.1200/OP.22.00315	Wed, 21 Sep 2022 06:00:00 -0400
38	pubmed:36130185	The role of microRNAs in the pathophysiology, diagnosis, and treatment of diabetic cardiomyopathy	Mahasin Abdel Rhman Peter Owira	CONCLUSION: MiRNAs serve as attractive potential targets for DCM diagnosis, prognosis and treatment due to their distinctive expression profile in DCM development.	pmid:36130185 doi:10.1093/jpp/rgac066	Wed, 21 Sep 2022 06:00:00 -0400
39	pubmed:36130290	Longitudinal study of patients with antimelanoma differentiation-associated gene 5 antibody-positive dermatomyositis associated interstitial lung disease	Xinyue Lian Yan Ye Jing Zou Chunmei Wu Shuang Ye Qiang Guo Sheng Chen Liangjing Lu Ran Wang Qiong Fu Chunde Bao	CONCLUSIONS: MDA5+ DM-ILD patients had a high mortality rate despite aggressive treatment. Patients who survived the first year usually showed a significant improvement in serological markers and pulmonary function during the long-term follow-up.	pmid:36130290 doi:10.1093/rheumatology/keac525	Wed, 21 Sep 2022 06:00:00 -0400