gene therapy

	NCT Number	Title	Authors	Description	Identifier	Dates
1	pubmed:36098613	Adolescent Idiopathic Scoliosis for Pediatric Providers	Connor L Zale Amy L McIntosh	Adolescent idiopathic scoliosis occurs in 2% to 3% of the adolescent population, affecting people between ages 10 and 16 years. Scoliosis onset is usually earlier in girls than in boys-generally occurring between ages 10 and 14 years for girls and 12 and 16 years for boys. The cause of idiopathic scoliosis is unknown. It is probably multifactorial. Much of the research on the etiology has focused on connective tissue abnormalities, nutritional deficiency, and genetic factors. [Pediatr Ann....	pmid:36098613 doi:10.3928/19382359-20220724-01	Tue, 13 Sep 2022 06:00:00 -0400
2	pubmed:36099255	Minimum inhibitory concentrations of rifampin and isoniazid among multidrug and isoniazid resistant Mycobacterium tuberculosis in Ethiopia	Muluwork Getahun Henry M Blumberg Gobena Ameni Dereje Beyene Russell R Kempker	CONCLUSION: Our findings suggest a role for rifabutin treatment in a subset of RR TB patients, thus potentially preserving an important drug class. The high proportion of moderate level INH resistant among MDR Mtb isolates indicates the potential benefit of high dose isoniazid treatment in a high proportion of katG gene harboring MDR Mtb isolates.	pmid:36099255 pmc:PMC9469996 doi:10.1371/journal.pone.0274426	Tue, 13 Sep 2022 06:00:00 -0400
3	pubmed:36100328	CRISPR/Cas9 gene editing: New hope for Alzheimer's disease therapeutics	Shanu Bhardwaj Kavindra Kumar Kesari Mahesh Rachamalla Shalini Mani Ghulam Md Ashraf Saurabh Kumar Jha Pravir Kumar Rashmi K Ambasta Harish Dureja Hari Prasad Devkota Gaurav Gupta Dinesh Kumar Chellappan Sachin Kumar Singh Kamal Dua Janne Ruokolainen Mohammad Amjad Kamal Shreesh Ojha Niraj Kumar Jha	BACKGROUND: Alzheimer's disease (AD) is an insidious, irreversible, and progressive neurodegenerative health condition manifesting as cognitive deficits and amyloid beta (A) plaques and neurofibrillary tangles. Approximately 50 million individuals are affected by AD, and the number is rapidly increasing globally. This review explores the role of CRISPR/Cas9 gene editing in the management of AD and its clinical manifestations.	pmid:36100328 doi:10.1016/j.jare.2021.07.001	Tue, 13 Sep 2022 06:00:00 -0400
4	pubmed:36100402	Matrix metalloproteinase 14 and plasma kallikrein 1 may be potential biomarkers in the diagnosis and treatment of sepsis: a proteomics and bioinformatics analysis	Libo Feng Yu Liu Muhu Chen Yingchun Hu	CONCLUSIONS: MMP14 and KLKB1 may be potential biomarkers for the diagnosis, treatment and prognosis of sepsis.	pmid:36100402 doi:10.3760/cma.j.cn121430-20210706-01011	Tue, 13 Sep 2022 06:00:00 -0400
5	pubmed:36100877	ZNF384-ZEB1 feedback loop regulates breast cancer metastasis	Qing-Xiang Meng Ke-Nie Wang Jun-Hui Li Hui Zhang Zhao-Hui Chen Xue-Jie Zhou Xu-Chen Cao Ping Wang Yue Yu	CONCLUSIONS: The findings suggest that ZNF384 can serve as a prognostic factor and a therapeutic target for breast cancer patients.	pmid:36100877 doi:10.1186/s10020-022-00541-1	Tue, 13 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
6	pubmed:36100891	Immune checkpoint inhibitor (ICI) genes and aging in malignant melanoma patients: a clinicogenomic TCGA study	Mohammed Safi Chenxing Jin Abdullah Aldanakh Ping Feng Henan Qin Mohammed Alradhi Lizhi Zhang Junying Zhang Salah Adlat Yi Zhao Jiwei Liu	CONCLUSIONS: Along with the risk score evaluation, the ICI gene (TNFRSF4) was identified as a tumor suppressor gene related to inequalities in age survival and associated with immune cell infiltrations. The aging responses of melanoma patients and related gene expression need further investigation in order to identify potential therapeutic targets.	pmid:36100891 doi:10.1186/s12885-022-09860-2	Tue, 13 Sep 2022 06:00:00 -0400
7	pubmed:36100894	KCNJ14 knockdown significantly inhibited the proliferation and migration of colorectal cells	Bin Li Ning Ge Zhongping Pan Chaofeng Hou Kun Xie Dongfang Wang Junwei Liu Jie Wan Feihong Deng Mengyi Li Shuping Luo	CONCLUSIONS: This study not only increases the molecular understanding of KCNJ14 but also provides a potentially valuable biological target for the treatment of colorectal cancer.	pmid:36100894 doi:10.1186/s12920-022-01351-4	Tue, 13 Sep 2022 06:00:00 -0400
8	pubmed:36101743	Integrated Analysis of the lncRNA-Associated ceRNA Network in Wilms Tumor via TARGET and GEO Databases	Biao An Yuan Hu Xiao Liang	Wilms tumor (WT) is the most common genitourinary renal tumor that typically occurs in children under 15 and is thought to be linked to somatic and germline mutations. However, the specific functional role of competing endogenous RNAs (ceRNAs) and their potential implications in WT remain unclear. In this study, we developed an lncRNA-mediated (long noncoding RNA-mediated) ceRNA network via the R packages for WT with expression data obtained from the tumor alterations relevant for...	pmid:36101743 pmc:PMC9452976 doi:10.1155/2022/2365991	Wed, 14 Sep 2022 06:00:00 -0400
9	pubmed:36103817	SerpinB3 drives cancer stem cell survival in glioblastoma	Adam Lauko Josephine Volovetz Soumya M Turaga Defne Bayik Daniel J Silver Kelly Mitchell Erin E Mulkearns-Hubert Dionysios C Watson Kiran Desai Manav Midha Jing Hao Kathleen McCortney Alicia Steffens Ulhas Naik Manmeet S Ahluwalia Shideng Bao Craig Horbinski Jennifer S Yu Justin D Lathia	Despite therapeutic interventions for glioblastoma (GBM), cancer stem cells (CSCs) drive recurrence. The precise mechanisms underlying CSC resistance, namely inhibition of cell death, are unclear. We built on previous observations that the high cell surface expression of junctional adhesion molecule-A drives CSC maintenance and identified downstream signaling networks, including the cysteine protease inhibitor SerpinB3. Using genetic depletion approaches, we found that SerpinB3 is necessary for...	pmid:36103817 doi:10.1016/j.celrep.2022.111348	Wed, 14 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
10	pubmed:36103832	Restoration of mitochondria axonal transport by adaptor Disc1 supplementation prevents neurodegeneration and rescues visual function	Heberto Quintero Yukihiro Shiga Nicolas Belforte Luis Alarcon-Martinez Sana El Hajji Deborah Villafranca-Baughman Florence Dotigny Adriana Di Polo	Deficits in mitochondrial transport are a common feature of neurodegenerative diseases. We investigated whether loss of components of the mitochondrial transport machinery impinge directly on metabolic stress, neuronal death, and circuit dysfunction. Using multiphoton microscope live imaging, we showed that ocular hypertension, a major risk factor in glaucoma, disrupts mitochondria anterograde axonal transport leading to energy decline in vulnerable neurons. Gene- and protein-expression analysis...	pmid:36103832 doi:10.1016/j.celrep.2022.111324	Wed, 14 Sep 2022 06:00:00 -0400
11	pubmed:36103843	Gene therapy for inherited retinal disease: long-term durability of effect	Bart P Leroy M Dominik Fischer John G Flannery Robert E MacLaren Deniz Dalkara Hendrik P N Scholl Daniel C Chung Claudio Spera Daniel Viriato Judit Banhazi	The recent approval of voretigene neparvovec (Luxturna®) for patients with biallelic RPE65 mutation-associated inherited retinal dystrophy with viable retinal cells represents an important step in the development of ocular gene therapies. Herein, we review studies investigating the episomal persistence of different recombinant adeno-associated virus (rAAV) vector genomes and the pre-clinical and clinical evidence of long-term effects of different RPE65 gene replacement therapies. A targeted...	pmid:36103843 doi:10.1159/000526317	Wed, 14 Sep 2022 06:00:00 -0400
12	pubmed:36103897	Adult Spinal Muscular Atrophy	Maggie C Walter Miriam Hiebeler	5q spinal muscular atrophy (SMA) is an autosomal recessive motor neuron disease affecting 1: 11000 live births and ranging from intrauterine to early adult onset. The course of the disease is progressive, the phenotype varies within a disease continuum and is mainly determined by the SMN2 copy number. So far, three disease modifying treatments (Nusinersen/Spinraza, Onasemnogene abeparvovec/Zolgensma, Risdiplam/Evrysdi) have been approved; however, gene replacement therapy with Onasemnogen...	pmid:36103897 doi:10.1055/a-1801-3785	Wed, 14 Sep 2022 06:00:00 -0400
13	pubmed:36103902	Are the epigenetic changes predictive of therapeutic efficacy for psychiatric disorders? A translational approach towards novel drug targets	Vincenzo Micale Martina Di Bartolomeo Serena Di Martino Tibor Stark Bernardo Dell'Osso Filippo Drago Claudio D'Addario	Although the etiopathogenesis of mental disorders is not fully understood, accumulating evidence support that clinical symptomatology cannot be assigned to a single gene mutation, but it involves several genetic factors. More specifically, a tight association between genes and environmental risk factors, which could be mediated by epigenetic mechanisms, may play a role in the development of mental disorders. Several data suggest that epigenetic modifications such as DNA methylation,...	pmid:36103902 doi:10.1016/j.pharmthera.2022.108279	Wed, 14 Sep 2022 06:00:00 -0400
14	pubmed:36103960	Anatabine attenuates ovalbumin-induced asthma via oxidative stress and inflammation mitigation and Nrf2/HO-1 signaling upregulation in rats	Walied Abdo Imer Haziri Mohamed Dmerdash Sulaiman Mohammed Alnasser Ali Hakamy Ehab Ali Soha A Soliman Hanan H Abd-Elhafeez Ahmed M Abd-Eldayem	AIMS: Asthma affects a large number of people worldwide and is characterized by chronic allergic airway inflammation. Anatabine is a natural alkaloid that is structurally similar to nicotine and found in the Solanaceae family of plants, with anti-inflammatory properties. Consequently, this study aimed to evaluate the potential therapeutic effect of anatabine against asthma.	pmid:36103960 doi:10.1016/j.lfs.2022.120954	Wed, 14 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
15	pubmed:36103998	Adeno-associated Virus Gene Therapy for Hemophilia	Benjamin J Samelson-Jones Lindsey A George	In vivo gene therapy is rapidly emerging as a new therapeutic paradigm for monogenic disorders. For almost three decades, hemophilia A (HA) and hemophilia B (HB) have served as model disorders for the development of gene therapy. This effort is soon to bear fruit with completed pivotal adeno-associated viral (AAV) vector gene addition trials reporting encouraging results and regulatory approval widely anticipated in the near future for the current generation of HA and HB AAV vectors. Here we...	pmid:36103998 doi:10.1146/annurev-med-043021-033013	Wed, 14 Sep 2022 06:00:00 -0400
16	pubmed:36104100	Restoration of p53 activity via intracellular protein delivery sensitizes triple negative breast cancer to anti-PD-1 immunotherapy	Zaofeng Yang Jacquelyne Ka-Li Sun Marianne M Lee Michael K Chan	CONCLUSION: This study validates that p53 restoration can be an effective approach to overcome ICI resistance and demonstrates that intracellular delivery of p53 protein can be an efficient, safe and potentially universal strategy to restore p53 activity in tumors carrying TP53 mutation.	pmid:36104100 doi:10.1136/jitc-2022-005068	Wed, 14 Sep 2022 06:00:00 -0400
17	pubmed:36104258	The effects of early exercise in traumatic brain-injured rats with changes in motor ability, brain tissue, and biomarkers	Chung Kwon Kim Jee Soo Park Eunji Kim Min-Kyun Oh Yong-Taek Lee Kyung Jae Yoon Kyeung Min Joo Kyunghoon Lee Young Sook Park	Traumatic brain injury (TBI) is brain damage which is caused by the impact of external mechanical forces. TBI can lead to the temporary or permanent impairment of physical and cognitive abilities, resulting in abnormal behavior. We recently observed that a single session of early exercise in animals with TBI improved their behavioral performance in the absence of other cognitive abnormalities. In the present study, we investigated the therapeutic effects of continuous exercise during the early...	pmid:36104258	Wed, 14 Sep 2022 06:00:00 -0400
18	pubmed:36104354	Epigenetic activation of the FLT3 gene by ZNF384 fusion confers a therapeutic susceptibility in acute lymphoblastic leukemia	Xujie Zhao Ping Wang Jonathan D Diedrich Brandon Smart Noemi Reyes Satoshi Yoshimura Jingliao Zhang Wentao Yang Kelly Barnett Beisi Xu Zhenhua Li Xin Huang Jiyang Yu Kristine Crews Allen Eng Juh Yeoh Marina Konopleva Chia-Lin Wei Ching-Hon Pui Daniel Savic Jun J Yang	FLT3 is an attractive therapeutic target in acute lymphoblastic leukemia (ALL) but the mechanism for its activation in this cancer is incompletely understood. Profiling global gene expression in large ALL cohorts, we identify over-expression of FLT3 in ZNF384-rearranged ALL, consistently across cases harboring different fusion partners with ZNF384. Mechanistically, we discover an intergenic enhancer element at the FLT3 locus that is exclusively activated in ZNF384-rearranged ALL, with the...	pmid:36104354 doi:10.1038/s41467-022-33143-w	Wed, 14 Sep 2022 06:00:00 -0400
19	pubmed:36104584	Characterization of phenylalanine hydroxylase gene variants and analysis of genotype-phenotype correlation in patients with phenylalanine hydroxylase deficiency from Fujian Province, Southeastern China	Jinfu Zhou Yinglin Zeng Xiaolong Qiu Qingying Lin Weifeng Chen Jinying Luo Liangpu Xu	CONCLUSIONS: Our study identified a PAH variant spectrum in PAHD patients from Fujian Province, Southeastern China. Quantitative correlation analysis between genotype and phenotype severity is helpful for genetic counseling and management.	pmid:36104584 doi:10.1007/s11033-022-07579-8	Wed, 14 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
20	pubmed:36104717	Naturally derived indole alkaloids targeting regulated cell death (RCD) for cancer therapy: from molecular mechanisms to potential therapeutic targets	Rui Qin Feng-Ming You Qian Zhao Xin Xie Cheng Peng Gu Zhan Bo Han	Regulated cell death (RCD) is a critical and active process that is controlled by specific signal transduction pathways and can be regulated by genetic signals or drug interventions. Meanwhile, RCD is closely related to the occurrence and therapy of multiple human cancers. Generally, RCD subroutines are the key signals of tumorigenesis, which are contributed to our better understanding of cancer pathogenesis and therapeutics. Indole alkaloids derived from natural sources are well defined for...	pmid:36104717 doi:10.1186/s13045-022-01350-z	Wed, 14 Sep 2022 06:00:00 -0400
21	pubmed:36104756	Human umbilical cord-derived mesenchymal stem cells ameliorate experimental colitis by normalizing the gut microbiota	Fan Yang Beibei Ni Qiuli Liu Fangping He Li Li Xuemei Zhong Xiaofan Zheng Jianxi Lu Xiaoyan Chen Huizhu Lin Ruixuan Xu Yizhan He Qi Zhang Xiaoguang Zou Wenjie Chen	BACKGROUND: Crohn's disease (CD) is a chronic non-specific inflammatory bowel disease. Current CD therapeutics cannot fundamentally change the natural course of CD. Therefore, it is of great significance to find new treatment strategies for CD. Preclinical and clinical studies have shown that mesenchymal stromal cells (MSCs) are a promising therapeutic approach. However, the mechanism by which MSCs alleviate CD and how MSCs affect gut microbes are still unclear and need further elucidation.	pmid:36104756 doi:10.1186/s13287-022-03118-1	Wed, 14 Sep 2022 06:00:00 -0400
22	pubmed:36104795	Elraglusib (9-ING-41), a selective small-molecule inhibitor of glycogen synthase kinase-3 beta, reduces expression of immune checkpoint molecules PD-1, TIGIT and LAG-3 and enhances $CD8^{+}$ T cell cytolytic killing of melanoma cells	Gary Shaw Ludimila Cavalcante Francis J Giles Alison Taylor	CONCLUSIONS: These data highlight the potential of elraglusib as an immune-modulatory agent and demonstrate the benefit of a sequential approach with immune checkpoint inhibition followed by GSK-3 inhibition in melanoma and provide a rationale for clinical investigation of elraglusib combined with immune checkpoint inhibitory molecules, including those targeting PD-1, TIGIT and LAG-3. This has several potential implications for current immunotherapy regimes, including possibly reducing the...	pmid:36104795 doi:10.1186/s13045-022-01352-x	Wed, 14 Sep 2022 06:00:00 -0400
23	pubmed:36104799	Characterization and management of facial angiofibroma related to tuberous sclerosis complex in the United States: retrospective analysis of the natural history database	Sreedevi Boggarapu Steven L Roberds JoAnne Nakagawa Eric Beresford	CONCLUSIONS: The presence of TSC2 mutations and most other TSC-related manifestations was significantly higher in patients with facial angiofibroma. About one-fourth of patients with facial angiofibroma used a topical mTOR inhibitor and use of systemic mTOR inhibitor for the management of facial angiofibroma or for the other manifestations was noted for 30.0%. About 44.6% of patients did not receive any treatment for the management of facial angiofibroma.	pmid:36104799 doi:10.1186/s13023-022-02496-2	Wed, 14 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
24	pubmed:36105076	FGF/FGFR-related lncRNAs based classification predicts prognosis and guides therapy in gastric cancer	Qiuxiang Chen Xiaojing Du	Fibroblast growth factor (FGF) and its receptor (FGFR) play crucial roles in gastric cancer (GC). Long non-coding RNAs (lncRNAs) are defined as RNA molecules of around 200 nucleotides or more, which are not translated into proteins. As well-known regulatory factors, lncRNAs are considered as biomarkers for prognosis and treatment response in GC. It is of importance to identify FGF/FGFR-related lncRNAs in GC. Here, some FGF/FGFR-related lncRNAs were identified in GC based on the data from public...	pmid:36105076 pmc:PMC9465033 doi:10.3389/fgene.2022.948102	Thu, 15 Sep 2022 06:00:00 -0400
25	pubmed:36105094	CENP-A is a potential prognostic biomarker and correlated with immune infiltration levels in glioma patients	Yuan Yang Mengyun Duan Yunfei Zha Zijun Wu	Background: Centromeric protein A (CENP-A), an essential protein involved in chromosomal segregation during cell division, is associated with several cancer types. However, its role in gliomas remains unclear. This study examined the clinical and prognostic significance of CENP-A in gliomas. Methods: Data of patients with glioma were collected from the Cancer Genome Atlas. Logistic regression, the Kruskal-Wallis test, and the Wilcoxon signed-rank test were performed to assess the relationship...	pmid:36105094 pmc:PMC9465177 doi:10.3389/fgene.2022.931222	Thu, 15 Sep 2022 06:00:00 -0400
26	pubmed:36105099	miR-22-3p as a potential biomarker for coronary artery disease based on integrated bioinformatics analysis	Minghua Zhang Yan Hu Haoda Li Xiaozi Guo Junhui Zhong Sha He	Background: Coronary artery disease (CAD) is a common cardiovascular disease that has attracted attention worldwide due to its high morbidity and mortality. Recent studies have shown that abnormal microRNA (miRNA) expression is effective in CAD diagnoses and processes. However, the potential relationship between miRNAs and CAD remains unclear. Methods: Microarray datasets GSE105449 and GSE28858 were downloaded directly from the Gene Expression Omnibus (GEO) to identify miRNAs involved in CAD....	pmid:36105099 pmc:PMC9464939 doi:10.3389/fgene.2022.936937	Thu, 15 Sep 2022 06:00:00 -0400
27	pubmed:36105107	Identification of hub genes related to CD4^{+} memory T cell infiltration with gene co-expression network predicts prognosis and immunotherapy effect in colon adenocarcinoma	Lingxue Tang Sheng Yu Qianqian Zhang Yinlian Cai Wen Li Senbang Yao Huaidong Cheng	Background: CD4^(+) memory T cells (CD4^(+) MTCs), as an important part of the microenvironment affecting tumorigenesis and progression, have rarely been systematically analyzed. Our purpose was to comprehensively analyze the effect of CD4^(+) MTC infiltration on the prognosis of colon adenocarcinoma (COAD). Methods: Based on RNA-Seq data, weighted gene co-expression network analysis (WGCNA) was used to screen the CD4^(+) MTC infiltration genes most associated with colon cancer and then identify...	pmid:36105107 pmc:PMC9465611 doi:10.3389/fgene.2022.915282	Thu, 15 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
28	pubmed:36105112	CDK2AP1 influences immune infiltrates and serves as a prognostic indicator for hepatocellular carcinoma	Yibin Che Ge Wang Qiang Xia	Background: Hepatocellular carcinoma (HCC) is a tumor with high malignancy and poor 5-years survival rate. Excellent tumor markers are very important for early clinical diagnosis and prognosis evaluation. Previous studies have shown that CDK2AP1 (Cyclin-dependent kinase 2-associated protein 1) is involved in cell-cycle and epigenetic regulation. In the present study, we assess CDK2AP1 expression, prognostic value, immunomodulatory and possible influencing pathways in HCC. Method: The Cancer...	pmid:36105112 pmc:PMC9465009 doi:10.3389/fgene.2022.937310	Thu, 15 Sep 2022 06:00:00 -0400
29	pubmed:36105184	Effect of theaflavin-3,3'-digallate on leptin-deficient induced nonalcoholic fatty liver disease might be related to lipid metabolism regulated by the Fads1/PPAR/Fabp4 axis and gut microbiota	Cheng Zhou Wenji Zhang Hui Lin Luyun Zhang Fan Wu Yan Wang Susu Yu Xinyue Peng Wenli Cheng Min Li Xiaoying Pan Zhenrui Huang Wenjuan Zhang	Nonalcoholic fatty liver disease (NAFLD), one of the risk factors for hepatitis, cirrhosis, and even hepatic carcinoma, has been a global public health problem. The polyphenol compound theaflavin-3,3'-digallate (TF3), mainly extracted from black tea, has been reported to produce an effect on hypoglycemic and antilipid deposition in vitro. In our study, we further investigated the function and novel mechanisms of TF3 in protecting NAFLD in vivo. By using leptin-deficient obese (ob/ob) mice with...	pmid:36105184 pmc:PMC9464872 doi:10.3389/fphar.2022.925264	Thu, 15 Sep 2022 06:00:00 -0400
30	pubmed:36105203	S-3'-hydroxy-7', 2', 4'-trimethoxyisoxane, a novel ferroptosis inducer, promotes NSCLC cell death through inhibiting Nrf2/HO-1 signaling pathway	Jing Chen Songlin Zhou Xian Zhang Huange Zhao	Background: Ferroptosis is a newly discovered and promising non-apoptotic programmed cell death (PCD), and inducing ferroptosis in cancer cells could open up a novel avenue for drug screening and cancer therapy. S-3'-hydroxy-7', 2', 4'-trimethoxyisoxane (ShtIX), a new isoflavane compound, has been reported to possess cytotoxicity in non-small cell lung cancer (NSCLC). The aim of this research is to explore the ShtIX-induced cell death form and its underlying molecular mechanism in NSCLC cells....	pmid:36105203 pmc:PMC9465255 doi:10.3389/fphar.2022.973611	Thu, 15 Sep 2022 06:00:00 -0400
31	pubmed:36105439	Clinical Characteristics and Gene Mutation Analysis of Poststroke Epilepsy	Deju Shen Yuqin Deng Chunyan Lin Jianshu Li Xuehua Lin Chaoning Zou	Epilepsy is one of the most common brain disorders worldwide. Poststroke epilepsy (PSE) affects functional retrieval after stroke and brings considerable social values. A stroke occurs when the blood circulation to the brain fails, causing speech difficulties, memory loss, and paralysis. An electroencephalogram (EEG) is a tool that may detect anomalies in brain electrical activity, including those induced by a stroke. Using EEG data to determine the electrical action in the brains of stroke...	pmid:36105439 pmc:PMC9444425 doi:10.1155/2022/4801037	Thu, 15 Sep 2022 06:00:00 -0400
32	pubmed:36105485	Identification of Warning Transition Points from Hepatitis B to Hepatocellular Carcinoma Based on Mutation Accumulation for the Early Diagnosis and Potential Drug Treatment of HBV-HCC	Fei Xu Qingkang Meng Feng Wu Yakun Wang Wenjun Yang Yun Tong Lei Liu Xiujie Chen	The accumulation of multiple genetic mutations is essential during the occurrence and development of hepatocellular carcinoma induced by hepatitis B (HBV-HCC), but understanding their cooperative effects and identifying the warning transition point from hepatitis B to HCC are challenges. In the genomic analysis of somatic mutations of the patient with HBV-HCC in a patient-specific protein-protein interaction (ps-PPI) network, we find mutation influence can propagate along the ps-PPI network....	pmid:36105485 pmc:PMC9467738 doi:10.1155/2022/3472179	Thu, 15 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
33	pubmed:36105536	The diagnostic significance of integrating m6A modification and immune microenvironment features based on bioinformatic investigation in aortic dissection	Ruiming Guo Jia Dai Hao Xu Suhua Zang Liang Zhang Ning Ma Xin Zhang Lixuan Zhao Hong Luo Donghai Liu Jian Zhang	CONCLUSION: This study indicated that FTO and IGF2BP1 were involved in the IME of AD. Integrating FTO and IGF2BP1 and MAPK1IP1L key genes in AD with a high m6A level context would provide clues for forthcoming AD diagnosis and therapy.	pmid:36105536 pmc:PMC9464924 doi:10.3389/fcvm.2022.948002	Thu, 15 Sep 2022 06:00:00 -0400
34	pubmed:36105586	Partial prevention of glucocorticoid-induced osteocyte deterioration in young male mice with osteocrin gene therapy	Courtney M Mazur Christian D Castro Andrade Nicha Tokavanich Tadatoshi Sato Michael Bruce Daniel J Brooks Mary L Bouxsein Jialiang S Wang Marc N Wein	Glucocorticoid excess suppresses osteocyte remodeling of surrounding bone minerals, causes apoptosis of osteoblasts and osteocytes, and disrupts bone remodeling, eventually, leading to glucocorticoid-induced osteoporosis and bone fragility. Preventing apoptosis and preserving osteocyte morphology could be an effective means of preventing bone loss during glucocorticoid treatment. We hypothesized that osteocrin, which preserves osteocyte viability and morphology in Sp7-deficient mice, could...	pmid:36105586 pmc:PMC9464962 doi:10.1016/j.isci.2022.105019	Thu, 15 Sep 2022 06:00:00 -0400
35	pubmed:36105615	Targeted Nanobubbles of PD-L1 mAb Combined with Doxorubicin as a Synergistic Tumor Repressor in Hepatocarcinoma	Yezi Chen Xiaoqin Luo Yun Liu Yunlei Zou Shiqi Yang Chaoqi Liu Yun Zhao	CONCLUSION: In summary, ultrasound-mediated PD-L1 mAb/DOX-NBs showed significant synergistic antitumor effects, providing a potential combined immunotherapy strategy for HCC.	pmid:36105615 pmc:PMC9464779 doi:10.2147/IJN.S376172	Thu, 15 Sep 2022 06:00:00 -0400
36	pubmed:36105674	Roles and current applications of S-nitrosoglutathione in anti-infective biomaterials	Hu Qian Zhimin Ye Lanping Pi Jun Ao	Bacterial infections can compromise the physical and biological functionalities of humans and pose a huge economical and psychological burden on infected patients. Nitric oxide (NO) is a broad-spectrum antimicrobial agent, whose mechanism of action is not affected by bacterial resistance. S-nitrosoglutathione (GSNO), an endogenous donor and carrier of NO, has gained increasing attention because of its potent antibacterial activity and efficient biocompatibility. Significant breakthroughs have...	pmid:36105674 pmc:PMC9465324 doi:10.1016/j.mtbio.2022.100419	Thu, 15 Sep 2022 06:00:00 -0400
37	pubmed:36105799	Investigation of the risk factors to predict cytokine release syndrome in relapsed or refractory B-cell acute lymphoblastic leukemia patients receiving IL-6 knocking down anti-CD19 chimeric antigen receptor T-cell therapy	Wen-Jie Gong Yan Qiu Ming-Hao Li Li-Yun Chen Yan-Yan Li Jing-Qiu Yu Li-Qing Kang Ai-Ning Sun De-Pei Wu Lei Yu Sheng-Li Xue	CD19 chimeric antigen receptor-T (CAR-T) cell therapy has achieved remarkable results in patients with relapsed or refractory B-cell acute lymphoblastic leukemia (r/r B-ALL). However, the cytokine release syndrome (CRS) was presented in most patients as common toxicity and severe CRS (sCRS) characterized by the sharp increase in interleukin-6 (IL-6) could be life-threatening. We conducted a phase II clinical trial of ssCAR-T-19 cells, anti-CD19 CAR-T cells with shRNA targeting IL-6, in 61...	pmid:36105799 pmc:PMC9464804 doi:10.3389/fimmu.2022.922212	Thu, 15 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
38	pubmed:36105853	Phenotype Analysis of Fused in Sarcoma Mutations in Amyotrophic Lateral Sclerosis	Maurizio Grassano Giorgia Brodini Giovanni De Marco Federico Casale Giuseppe Fuda Paolina Salamone Maura Brunetti Luca Sbaiz Salvatore Gallone Paolo Cugnasco Alessandro Bombaci Rosario Vasta Umberto Manera Antonio Canosa Cristina Moglia Andrea Calvo Bryan J Traynor Adriano Chio	BACKGROUND AND OBJECTIVES: Pathogenic variations in fused in sarcoma (FUS) are among the most common genetic causes of amyotrophic lateral sclerosis (ALS) worldwide. They are supposedly characterized by a homogeneous pure motor phenotype with early-onset and short disease duration. However, a few FUS-mutated cases with a very late disease onset and slow progression have been reported. To analyze genotype-phenotype correlations and identify the prognostic factors in FUS-ALS cases.	pmid:36105853 pmc:PMC9469212 doi:10.1212/NXG.0000000000200011	Thu, 15 Sep 2022 06:00:00 -0400
39	pubmed:36106113	Adjuvant crizotinib in high-risk uveal melanoma following definitive therapy	Shaheer Khan Jose Lutzky Alexander N Shoushtari Joanne Jeter Brian Marr Thomas E Olencki Colleen M Cebulla Mohamed Abdel-Rahman J William Harbour Naomi Sender Alexandra Nesson Shahnaz Singh-Kandah Susana Hernandez Jeanelle King Manpreet S Katari Lyssa Dimapanat Stephanie Izard Grazia Ambrosini Oliver Surriga Alex J Rai Codruta Chiuzan Gary K Schwartz Richard D Carvajal	CONCLUSIONS: The use of adjuvant crizotinib in patients with high-risk UM did not result in improved RFS when compared to historical controls. Analysis of blood extracellular vesicles revealed changes in protein content associated with treatment, raising the possibility of future use as a biomarker. Further investigation of adjuvant treatment options are necessary for this challenging disease.	pmid:36106113 pmc:PMC9465386 doi:10.3389/fonc.2022.976837	Thu, 15 Sep 2022 06:00:00 -0400
40	pubmed:36106115	Enhancing anti-tumour innate immunity by targeting the DNA damage response and pattern recognition receptors in combination with radiotherapy	Charleen M L Chan Wah Hak Antonio Rullan Emmanuel C Patin Malin Pedersen Alan A Melcher Kevin J Harrington	Radiotherapy is one of the most effective and frequently used treatments for a wide range of cancers. In addition to its direct anti-cancer cytotoxic effects, ionising radiation can augment the anti-tumour immune response by triggering pro-inflammatory signals, DNA damage-induced immunogenic cell death and innate immune activation. Anti-tumour innate immunity can result from recruitment and stimulation of dendritic cells (DCs) which leads to tumour-specific adaptive T-cell priming and...	pmid:36106115 pmc:PMC9465159 doi:10.3389/fonc.2022.971959	Thu, 15 Sep 2022 06:00:00 -0400
41	pubmed:36106123	A novel cuproptosis-related lncRNA nomogram to improve the prognosis prediction of gastric cancer	Anqi Feng Lingnan He Tao Chen Meidong Xu	CONCLUSION: A novel cuproptosis-related lncRNAs signature impacts on the prognosis and immunological features of GC.	pmid:36106123 pmc:PMC9465020 doi:10.3389/fonc.2022.957966	Thu, 15 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
42	pubmed:36106325	Molecular transmission network of pretreatment drug resistance among human immunodeficiency virus-positive individuals and the impact of virological failure on those who received antiretroviral therapy in China	Hongli Chen Jing Hu Chang Song Miaomiao Li Yesheng Zhou Aobo Dong Ruihua Kang Jingjing Hao Jiaxin Zhang Xiu Liu Dan Li Yi Feng Lingjie Liao Yuhua Ruan Hui Xing Yiming Shao	CONCLUSION: The overall prevalence of PDR was close to a high level and had an impact on virological failure after 3 years of ART. Moreover, HIV drug-resistant strains were transmitted in the molecular transmission network. These results illustrate the importance of monitoring PDR and ensuring virological suppression through drug adherence.	pmid:36106325 pmc:PMC9464856 doi:10.3389/fmed.2022.965836	Thu, 15 Sep 2022 06:00:00 -0400
43	pubmed:36106336	Importance of predicting non-response to intravenous immunoglobulin therapy in non-Asian patients with Kawasaki disease	Isabelle Koné-Paut Maryam Piram	No abstract	pmid:36106336 pmc:PMC9465312 doi:10.1016/j.lanepe.2022.100507	Thu, 15 Sep 2022 06:00:00 -0400
44	pubmed:36106376	Clinical Utility of Comprehensive Genomic Profiling Tests for Advanced or Metastatic Solid Tumor in Clinical Practice	Hanae Ida Takafumi Koyama Takaaki Mizuno Kuniko Sunami Takashi Kubo Kazuki Sudo Kayoko Tao Makoto Hirata Kan Yonemori Ken Kato Takuji Okusaka Yuichiro Ohe Yoshiyuki Matsui Naoya Yamazaki Chitose Ogawa Akira Kawai Yoshitaka Narita Minoru Esaki Noboru Yamamoto	Previous clinical trials indicate that 10% to 25% of patients received genomically matched therapy after the comprehensive genomic profiling (CGP) tests. However, the clinical utility of CGP tests has not been assessed in clinical practice. We assessed the clinical utility of CGP tests for advanced or metastatic solid tumor and determined the proportion of patients receiving genomically matched therapy among those with common and non-common cancers. From August 2019 to July 2020, a total of 418...	pmid:36106376 doi:10.1111/cas.15586	Thu, 15 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
45	pubmed:36106631	Increased p53 expression induced by APR-246 reprograms tumor-associated macrophages to augment immune checkpoint blockade	Arnab Ghosh Judith Michels Riccardo Mezzadra Divya Venkatesh Lauren Dong Ricardo Gomez Fadi Samaan Yu-Jui Ho Luis Felipe Campesato Levi Mangarin John Fak Nathan Suek Aliya Holland Cailian Liu Mohsen Abu-Akeel Yonina Bykov Hong Zhong Kelly Fitzgerald Sadna Budhu Andrew Chow Roberta Zappasodi Katherine S Panageas Olivier de Henau Marcus Ruscetti Scott W Lowe Taha Merghoub Jedd D Wolchok	In addition to playing a major role in tumor cell biology, p53 generates a microenvironment that promotes antitumor immune surveillance via tumor-associated macrophages. We examined whether increasing p53 signaling in the tumor microenvironment influences antitumor T cell immunity. Our findings indicate that increased p53 signaling induced either pharmacologically with APR-246 (eprenetapopt) or in p53-overexpressing transgenic mice can disinhibit antitumor T cell immunity and augment the...	pmid:36106631 doi:10.1172/JCI148141	Thu, 15 Sep 2022 06:00:00 -0400
46	pubmed:36106640	The FoxO4/DKK3 axis represses IFN- expression by Th1 cells and limits antimicrobial immunity	Xiang Chen Jia Hu Yunfei Wang Younghee Lee Xiaohong Zhao Huiping Lu Gengzhen Zhu Hui Wang Yu Jiang Fan Liu Yongzhen Chen Byung-Seok Kim Qinghua Zhou Xindong Liu Xiaohu Wang Seon Hee Chang Chen Dong	Forkhead box O transcriptional factors, especially FoxO1 and FoxO3a, play critical roles in physiologic and pathologic immune responses. However, the function of FoxO4, another main member of the FoxO family, in lymphoid cells is still poorly understood. Here, we showed that loss of FoxO4 in T cells augmented IFN- production of Th1 cells in vitro. Correspondingly, conditional deletion of FoxO4 in CD4+ T cells enhanced T cell-specific responses to Listeria monocytogenes infection in vivo....	pmid:36106640 doi:10.1172/JCI147566	Thu, 15 Sep 2022 06:00:00 -0400
47	pubmed:36106661	R-spondin-3 is an oncogenic driver of poorly differentiated invasive breast cancer	Eline J Ter Steege Mandy Boer Nikki C Timmer Carola Me Ammerlaan Ji-Ying Song Patrick Wb Derksen John Hilkens Elvira Rm Bakker	R-spondins (RSPOs) are influential signaling molecules that promote the Wnt/-catenin pathway and self-renewal of stem cells. Currently, RSPOs are emerging as clinically relevant oncogenes, being linked to cancer development in multiple organs. Although this has instigated the rapid development and testing of therapeutic antibodies targeting RSPOs, functional evidence that RSPO causally drives cancer has focused primarily on the intestinal tract. Here, we assess the oncogenic capacity of RSPO in...	pmid:36106661 doi:10.1002/path.5999	Thu, 15 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
48	pubmed:36106778	Prenatal Somatic Cell Gene Therapies: Charting a Path Toward Clinical Applications (Proceedings of the CERSI-FDA Meeting)	Akos Herzeg Graça Almeida-Porada R Alta Charo Anna L David Juan Gonzalez-Velez Nalin Gupta Larissa Lapteva Billie Lianoglou William Peranteau Christopher Porada Stephan J Sanders Teresa N Sparks David H Stitelman Evi Struble Charlotte J Sumner Tippi C MacKenzie	We are living in a golden age of medicine in which the availability of prenatal diagnosis, fetal therapy, and gene therapy/editing make it theoretically possible to repair almost any defect in the genetic code. Furthermore, the ability to diagnose genetic disorders before birth and the presence of established surgical techniques enable these therapies to be delivered safely to the fetus. Prenatal therapies are generally used in the second or early third trimester for severe, life-threatening...	pmid:36106778 doi:10.1002/jcph.2127	Thu, 15 Sep 2022 06:00:00 -0400
49	pubmed:36106796	Analysis for variable manifestations and molecular characteristics of pyridox(am)ine-5'-phosphate oxidase (PNPO) deficiency	Xianru Jiao Pan Gong Yue Niu Zhao Xu Yuehua Zhang Zhixian Yang	CONCLUSIONS: The clinical characteristics, including age of onset, treatment response, and prognosis, were variable and difficult to classify into different types clearly. Patients with PNPO deficiency who used PN as their main treatment and being able to control seizures seemed to be associated with better outcomes. Patients with the same genotype tended to show the correlation of phenotype-genotype.	pmid:36106796 doi:10.1093/hmg/ddac234	Thu, 15 Sep 2022 06:00:00 -0400
50	pubmed:36107131	Copper nanocrystalline-doped folic acid-based super carbon dots for an enhanced antitumor effect in response to tumor microenvironment stimuli	Qing Xia Ying Zhang Hui Zhang Xiong Zhang Xiaodan Wu Zhiqiang Wang Rui Yan Yingxue Jin	Chemodynamic therapy (CDT) is a promising cancer treatment strategy to induce tumor cell apoptosis with harmful reactive oxygen species (ROS), yet over-expression of glutathione (GSH) in the tumor microenvironment (TME) severely depletes the ROS and limits the CDT efficacy. Copper-containing materials could efficiently decrease the level of GSH in the TME. In this study, copper nanocrystalline-doped folic acid-based super carbon dots (FA-CDs@Cu^(x)) were prepared to realize an enhanced antitumor...	pmid:36107131 doi:10.1039/d2tb01363k	Thu, 15 Sep 2022 06:00:00 -0400
51	pubmed:36107182	Phage therapy of purulent-inflammatory pathology of the outer and middle ear	A V Gurov E G Lapenko A I Kryukov	The steady growth of antibiotic resistance of bacteria and, as a result, difficulties in selecting effective drugs determine the search for an alternative strategy for the use of antimicrobials, and therapy based on the use of bacteriophages is such. Phage therapy in otorhinolaryngology is actively used for the treatment of rhinosinusitis, diseases of the pharynx and larynx. However, it should be noted that the use of this group of drugs in the treatment of ear diseases is not sufficiently...	pmid:36107182 doi:10.17116/otorino20228704156	Thu, 15 Sep 2022 06:00:00 -0400
52	pubmed:36107200	Nanomedicine and drug delivery to the retina: current status and implications for gene therapy	Mohamed Tawfik Fang Chen Jeffrey L Goldberg Bernhard A Sabel	Blindness affects more than 60 million people worldwide. Retinal disorders, including age-related macular degeneration (AMD), diabetic retinopathy (DR), and glaucoma, are the leading causes of blindness. Finding means to optimize local and sustained delivery of drugs or genes to the eye and retina is one goal to advance the development of new therapeutics. Despite the ease of accessibility of delivering drugs via the ocular surface, the delivery of drugs to the retina is still challenging due to...	pmid:36107200 doi:10.1007/s00210-022-02287-3	Thu, 15 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
53	pubmed:36107226	Treatment of cerebral adrenoleukodystrophy: allogeneic transplantation and lentiviral gene therapy	Ashish O Gupta Gerald Raymond Rene I Pierpont Stephan Kemp R Scott McIvor Arpana Rayannavar Bradley Miller Troy C Lund Paul J Orchard	INTRODUCTION: Adrenoleukodystrophy (ALD) is an X-linked peroxisomal disorder with an incidence of 1 in 14-17,000 male births, caused by pathogenic variants within the ABCD1 gene. By adulthood, approximately 40% of patients develop cerebral ALD, a severe, neuroinflammatory condition that is generally progressive and fatal without intervention.	pmid:36107226 doi:10.1080/14712598.2022.2124857	Thu, 15 Sep 2022 06:00:00 -0400
54	pubmed:36107324	Prostate Cancer Epigenetic Plasticity and Enhancer Heterogeneity: Molecular Causes, Consequences and Clinical Implications	Jeroen Kneppers Andries M Bergman Wilbert Zwart	Prostate cancer (PCa) proliferation is dictated by androgen receptor (AR) signaling, which regulates gene expression through cis-regulatory regions including proximal and distal enhancers. The repertoire of AR interactions at enhancers is dependent on tissue and cellular contexts and thus shape a spectrum of phenotypes through such epigenetic heterogeneity. Moreover, PCa is a multifocal disease and displays a high degree of intra- and inter-tumor heterogeneity, adding to the phenotypic...	pmid:36107324 doi:10.1007/978-3-031-11836-4_15	Thu, 15 Sep 2022 06:00:00 -0400
55	pubmed:36107325	Epigenetic Coregulation of Androgen Receptor Signaling	Rayzel C Fernandes Damien A Leach Charlotte L Bevan	The androgen receptor (AR) is a ligand-activated transcription factor belonging to the nuclear receptor (NR) superfamily. As with other members of the NR family, transcriptional activity of the AR is regulated by interactions with coregulatory proteins, which either enhance (coactivators) or repress (corepressors) its transcriptional activity. AR associated coregulators are functionally diverse, but a large fraction are epigenetic histone and chromatin modifiers. Epigenetic coregulators are...	pmid:36107325 doi:10.1007/978-3-031-11836-4_16	Thu, 15 Sep 2022 06:00:00 -0400
56	pubmed:36107326	Clinical Translation: Targeting the Estrogen Receptor	Ciara Metcalfe Jennifer O Lauchle	Estrogen Receptor alpha (ER) stands as one of the most successfully prosecuted drug targets in oncology, beginning with the approval of tamoxifen for women with ER positive (ER+) breast cancer over 40 years ago. The field continued to advance with the development of aromatase inhibitors and the pure antiestrogen fulvestrant. With multiple endocrine therapies approved for the treatment of ER+ breast cancer, efforts to generate novel ER-targeted therapeutics somewhat diminished in the early...	pmid:36107326 doi:10.1007/978-3-031-11836-4_17	Thu, 15 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
57	pubmed:36107428	Comparison of Management and Outcomes in ERBB2-Low vs ERBB2-Zero Metastatic Breast Cancer in France	Ombline de Calbiac Amélie Lusque Audrey Mailliez Thomas Bachelot Lionel Uwer Marie-Ange Mouret-Reynier George Emile Christelle Jouannaud Anthony Gonçalves Anne Patsouris Véronique Diéras Marianne Leheurteur Thierry Petit Paul Cottu Jean-Marc Ferrero Véronique D'Hondt Isabelle Desmoulins Joana Mourato-Ribeiro Anne-Laure Martin Jean-Sébastien Frenel	CONCLUSIONS AND RELEVANCE: In this large cohort study, patients with ERBB2-low MBC had a slightly better OS than those with completely ERBB2-zero tumors, but identical PFS1, which could help guide treatment selection.	pmid:36107428 doi:10.1001/jamanetworkopen.2022.31170	Thu, 15 Sep 2022 06:00:00 -0400
58	pubmed:36107483	Individualizing the Oncological Treatment of Patients With Metastatic Non-Clear Cell Renal Cell Carcinoma by Using Gene Sequencing and Patient-Reported Outcomes: Protocol for the INDIGO Study	Ida Marie Lind Rasmussen Anne Vest Soerensen Anne Kirstine Møller Gitte Fredberg Persson Jesper Andreas Palshof Gry Assam Taarnhøj Helle Pappot	CONCLUSIONS: We aim to explore methods for improving the treatment outcomes of patients with non-CC RCC, and the INDIGO study will contribute further data on personalized medicine for rare types of RCC and provide new knowledge on the active use of electronic PROs.	pmid:36107483 doi:10.2196/36632	Thu, 15 Sep 2022 06:00:00 -0400
59	pubmed:36107502	Deciphering SARS CoV-2-associated pathways from RNA sequencing data of COVID-19-infected A549 cells and potential therapeutics using in silico methods	Peter Natesan Pushparaj Laila Abdullah Damiati Iuliana Denetiu Sherin Bakhashab Muhammad Asif Abrar Hussain Sagheer Ahmed Mohammad Hamid Hamdard Mahmood Rasool	CONCLUSIONS: In conclusion, we have used the in silico next-generation knowledge discovery (NGKD) methods to discover COVID-19-associated pathways and specific therapeutics that have the potential to ameliorate the disease pathologies associated with COVID-19.	pmid:36107502 doi:10.1097/MD.0000000000029554	Thu, 15 Sep 2022 06:00:00 -0400
60	pubmed:36107507	Lung adenocarcinoma with EGFR 19Del and an ALK rearrangement benefits from alectinib instead of an EGFR-TKI: A case report	Hongbiao Wang Sujuan Zhu Zhifeng Li Xiaofang Qi Liwen Zhang Leiyu Ke Yingcheng Lin	RATIONALE: A remarkable concurrence of an EGFR mutation and an EML4-ALK fusion (double positive) occasionally occurs within a narrow number of patients. Previous studies using targeted therapy on EGFR/ALK co-mutated patients have commonly focused on single tyrosine kinase inhibitors (TKIs) or on the sequential use of EGFR-TKIs and ALK-TKIs. At present, no consensus exists regarding the treatment of patients with double positive mutations. The effectiveness of precision therapy also remains...	pmid:36107507 doi:10.1097/MD.0000000000030316	Thu, 15 Sep 2022 06:00:00 -0400
61	pubmed:36107601	15-Year progression to liver cancer in the lack of treatment for lysosomal acid lipase deficiency: A case report	Marlone Cunha-Silva Eloy Vianey Carvalho de França Clauber Teles Veiga Raquel Dias Greca Priscilla Brito Sena de Moraes Daniel Ferraz de Campos Mazo Elaine Cristina de Ataíde Simone Reges Perales Leonardo Trevizan Monici Tiago Sevá-Pereira	RATIONALE: Lysosomal acid lipase deficiency (LAL-D) is a poorly diagnosed genetic disorder characterized by the accumulation of cholesteryl esters and triglycerides in many tissues, leading to dyslipidemia and cardiovascular complications. In the liver, deposits are found within hepatocytes and Kupffer cells, generating microvesicular steatosis, progressive fibrosis, and cirrhosis. Sebelipase alfa is the target therapy which can improve laboratory changes and reduce the progression of liver...	pmid:36107601 doi:10.1097/MD.0000000000030315	Thu, 15 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
62	pubmed:36107619	Keratinocyte-derived cytokine TSLP promotes growth and metastasis of melanoma by regulating the tumor-associated immune microenvironment	Wenjin Yao Beatriz German Dounia Chraa Antoine Braud Cecile Hugel Pierre Meyer Guillaume Davidson Patrick Laurette Gabrielle Mengus Eric Flatter Pierre Marschall Justine Segaud Marine Guivarch Pierre Hener Marie-Christine Birling Dan Lipsker Irwin Davidson Mei Li	Malignant melanoma is a major public health issue displaying frequent resistance to targeted therapy and immunotherapy. A major challenge is to better understand how melanoma cells evade immune elimination and how tumor growth and metastasis is facilitated by tumor microenvironment. Here, we show that expression of the cytokine TSLP by epidermal keratinocytes is induced by cutaneous melanoma in both mice and humans. Using genetically engineered models of melanoma and tumor cell grafting combined...	pmid:36107619 doi:10.1172/jci.insight.161438	Thu, 15 Sep 2022 06:00:00 -0400
63	pubmed:36107810	APPROACH TO THE PATIENT WITH CONGENITAL HYPOTHYROIDISM	Athanasia Stoupa Dulanjalee Kariyawasam Adrien Nguyen Quoc Michel Polak Aurore Carré	Congenital hypothyroidism (CH) is the most frequent neonatal endocrine disorder and the most common preventable cause of development delay and growth failure, if diagnosed and treated early. The thyroid is the first endocrine gland to develop during embryonic life and to be recognizable in humans. Thyroid development and maturation can be divided into 2 phases: a first phase of embryogenesis and a second phase of folliculogenesis and differentiation with thyroid hormone production at the final...	pmid:36107810 doi:10.1210/clinem/dgac534	Thu, 15 Sep 2022 06:00:00 -0400
64	pubmed:36108259	Genomic Classification of HER2-Positive Patients With 80-Gene and 70-Gene Signatures Identifies Diversity in Clinical Outcomes With HER2-Targeted Neoadjuvant Therapy	Pat W Whitworth Peter D Beitsch Mary K Murray Paul D Richards Angela Mislowsky Carrie L Dul James V Pellicane Paul L Baron Rakhshanda Layeequr Rahman Laura A Lee Beth B Dupree Pond R Kelemen Andrew Y Ashikari Raye J Budway Cristina Lopez-Penalver William Dooley Shiyu Wang Patricia Dauer Andrea R Menicucci Erin B Yoder Christine Finn Lisa E Blumencranz William Audeh	CONCLUSION: The 80-gene assay identified meaningful genomic diversity in patients with cHER2 disease. Patients with cHER2/gHER2 tumors, who benefitted most from dual HER2-targeted therapy, accounted for approximately half of the cHER2 cohort. Genomically Luminal tumors had low pCR rates but good 5-year outcomes. cHER2/gBasal tumors derived no benefit from dual therapy and had significantly worse 5-year prognosis; these patients merit special consideration in future trials.	pmid:36108259 doi:10.1200/PO.22.00197	Thu, 15 Sep 2022 06:00:00 -0400