gene therapy

	NCT Number	Title	Authors	Description	Identifier	Dates
1	pubmed:36054980	The short-term predictive value of CD4^{+} cells for combination therapy with high-dose dexamethasone and immunoglobulin in newly diagnosed primary immune thrombocytopenia patients	Hongyun Liu Xiaoyan Liu Guoyang Zhang Jieyu Wang Duolan Naren Shuangfeng Xie Yiqing Li Danian Nie Zhixiong Li Liping Ma	CONCLUSIONS: Our results indicate that Th1, Th17, and Treg cells and IL-2 and IL-23 participate in the onset of ITP. Higher profiles of Th2, IL-2 and IL-23 may predict poor treatment outcomes. Higher levels of IL-17 and lower profile of Treg may predict sensitivity to HD DXM and IVIg combination therapy.	pmid:36054980 doi:10.1016/j.thromres.2022.08.014	Fri, 02 Sep 2022 06:00:00 -0400
2	pubmed:36055049	Methylation subgroup and molecular heterogeneity is a hallmark of glioblastoma: implications for biopsy targeting, classification and therapy	J Gempt F Withake A K Aftahy H S Meyer M Barz C Delbridge F Liesche-Starnecker G Prokop N Pfarr J Schlegel B Meyer C Zimmer B H Menze B Wiestler	CONCLUSIONS: (Epi)genetic intratumoral heterogeneity is a hallmark of GB, both at DNA methylation and CNV level. This intratumoral heterogeneity is of utmost importance for molecular classification as well as for defining therapeutic targets in this disease, as single biopsies might underestimate the true molecular diversity in a tumor.	pmid:36055049 doi:10.1016/j.esmoop.2022.100566	Fri, 02 Sep 2022 06:00:00 -0400
3	pubmed:36055084	Immunological profile in a pediatric population of patients with spherocytosis. A single-center experience	Silvio Marchesani Letizia Sabatini Valentina Bertaina Olivia Marini Michela Ambrosi Margherita Di Mauro Matilde Cossutta Livia Schettini Mariachiara Lodi Gioacchino Andrea Rotulo Paolo Palma Giuseppe Palumbo Giulia Ceglie	Spherocytosis is a hereditary disease caused by the deficiencies of different membrane proteins of red blood cells. Currently, splenectomy is the main therapeutic strategy available, although it is accompanied by an increased risk of sepsis. Several evidences have supported the hypothesis of spleen dysfunction in patients with spherocytosis that haven't yet undergone splenectomy. The aim of this study is to furtherly characterize this aspect, by describing the immune subpopulations in peripheral...	pmid:36055084 doi:10.1016/j.bcmd.2022.102700	Fri, 02 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
4	pubmed:36055212	Identification and single-base gene-editing functional validation of a cis-EPO variant as a genetic predictor for EPO-increasing therapies	Charli E Harlow Josan Gandawijaya Rosemary A Bamford Emily-Rose Martin Andrew R Wood Peter J van der Most Toshiko Tanaka Hampton L Leonard Amy S Etheridge Federico Innocenti Robin N Beaumont Jessica Tyrrell Mike A Nalls Eleanor M Simonsick Pranav S Garimella Eric J Shiroma Niek Verweij Peter van der Meer Ron T Gansevoort Harold Snieder Paul J Gallins Dereje D Jima Fred Wright Yi-Hui Zhou Luigi Ferrucci Stefania Bandinelli Dena G Hernandez Pim van der Harst Vickas V Patel Dawn M Waterworth Audrey Y Chu Asami Oguro-Ando Timothy M Frayling	Hypoxia-inducible factor prolyl hydroxylase inhibitors (HIF-PHIs) are currently under clinical development for treating anemia in chronic kidney disease (CKD), but it is important to monitor their cardiovascular safety. Genetic variants can be used as predictors to help inform the potential risk of adverse effects associated with drug treatments. We therefore aimed to use human genetics to help assess the risk of adverse cardiovascular events associated with therapeutically altered EPO levels to...	pmid:36055212 doi:10.1016/j.ajhg.2022.08.004	Fri, 02 Sep 2022 06:00:00 -0400
5	pubmed:36055241	Whole-genome CRISPR screening identifies genetic manipulations to reduce immune rejection of stem cell-derived islets	Elad Sintov Igor Nikolskiy Victor Barrera Jennifer Hyoje-Ryu Kenty Alexander S Atkin Dario Gerace Shannan J Ho Sui Kyle Boulanger Douglas A Melton	Human embryonic stem cells (hESCs) provide opportunities for cell replacement therapy of insulin-dependent diabetes. Therapeutic quantities of human stem cell-derived islets (SC-islets) can be produced by directed differentiation. However, preventing allo-rejection and recurring autoimmunity, without the use of encapsulation or systemic immunosuppressants, remains a challenge. An attractive approach is to transplant SC-islets, genetically modified to reduce the impact of immune rejection. To...	pmid:36055241 doi:10.1016/j.stemcr.2022.08.002	Fri, 02 Sep 2022 06:00:00 -0400
6	pubmed:36055365	Non-coding RNAs in EMT regulation: Association with tumor progression and therapy response	Mehrdokht Sadrkhanloo Maliheh Entezari Mohsen Rashidi Mehrdad Hashemi Rasoul Raesi Sam Saghari Salman Daneshi Shokooh Salimimoghadam Kiavash Hushmandi Sepideh Mirzaei Afshin Taheriazam	RNA molecules lacking capacity in protein translation, are known as non-coding RNAs (ncRNAs). Growth, differentiation and migration are influenced by ncRNAs in cells. The abnormal expression of ncRNAs contributes to development of diseases, especially cancer. On the other hand, EMT is a vital mechanism for cancer invasion and diffusion in body. In this manuscript, role of ncRNAs in EMT regulation and subsequent effect on cancer progression is investigated. The miRNAs regulate EMT by affecting...	pmid:36055365 doi:10.1016/j.ejphar.2022.175212	Fri, 02 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
7	pubmed:36055405	Leukocyte cell-derived chemotaxin 2 regulates epithelial-mesenchymal transition and cancer stemness in hepatocellular carcinoma	Tian-Huei Chu Chou-Yuan Ko Po-Han Tai Yi-Chen Chang Chao-Cheng Huang Tung-Yang Wu Hoi-Hung Chan Ping-Hsuan Wu Chien-Hui Weng Yu-Wei Lin Mei-Lang Kung Cheng-Chieh Fang Jian-Ching Wu Zhi-Hong Wen Yung-Kuo Lee Tsung-Hui Hu Ming-Hong Tai	Leukocyte cell-derived chemotaxin 2 (LECT2) acts as a tumor suppressor in hepatocellular carcinoma (HCC). However, the anti-neoplastic mechanism of LECT2, especially its influence on hepatic cancer stem cells (CSCs), remains largely unknown. In The Cancer Genome Atlas (TCGA) cohort, LECT2 mRNA expression was shown to be associated with stage, grade, recurrence, and overall survival in human HCC patients, and LECT2 expression was downregulated in hepatoma tissues compared with the adjacent...	pmid:36055405 doi:10.1016/j.jbc.2022.102442	Fri, 02 Sep 2022 06:00:00 -0400
8	pubmed:36055553	Midterm outcomes of isolated thoracic aortic replacement in congenital versus degenerative aortopathy in a 15-year institutional cohort	Rebecca Sorber Lillian L Tsai Caitlin W Hicks James H Black	CONCLUSIONS: While carrying significant operative risks and potential for morbidity, open thoracic aortic replacement represents a well-tolerated, durable treatment option for patients with congenitally mediated thoracic aortic disease. Since both CTD and non-CTD patients who required thoracic aortic replacement often need future aortic intervention, vigilant surveillance is warranted. Equivalent intervention rates between the two groups suggest remodeling of the CTD aorta is almost universally...	pmid:36055553 doi:10.1016/j.jvs.2022.05.033	Fri, 02 Sep 2022 06:00:00 -0400
9	pubmed:36055605	Comparison of transcriptome profiles of mesenchymal stem cells derived from umbilical cord and bone marrow of giant panda (Ailuropoda melanoleuca)	Dong-Hui Wang Jia-Song Chen Rong Hou Yuan Li Jun-Hui An Ping He Zhi-Gang Cai Xiao-Hu Liang Yu-Liang Liu	Mesenchymal stem cells (MSCs) have pluripotent differentiation ability and play an important role in human clinical cell therapy. While, the research on MSCs in endangered wild animals is extremely rare. In our previous studies, the bone marrow mesenchymal stem cells (bmMSCs) and umbilical cord mesenchymal stem cells (ucMSCs) of giant panda (Ailuropoda melanoleuca) were successfully isolated. We aimed to characterize the differences in gene expression profiles between these two types of MSCs...	pmid:36055605 doi:10.1016/j.gene.2022.146854	Fri, 02 Sep 2022 06:00:00 -0400
10	pubmed:36055775	Multistage-Responsive Gene Editing to Sensitize Ion-Interference Enhanced Carbon Monoxide Gas Therapy	Yayao Li Yongchun Pan Chao Chen Zekun Li Shiyu Du Xiaowei Luan Yanfeng Gao Xin Han Yujun Song	As a promising therapeutic modality targeting cancer, gas therapy still faces critical challenges, especially in enhancing therapeutic efficacy and avoiding gas poisoning risks. Here, a pH/glutathione (GSH) dual stimuli-responsive CRISPR/Cas9 gene-editing nanoplatform combined with calcium-enhanced CO gas therapy for precise anticancer therapy, is established. In the tumor microenvironment (TME), the fast biodegradation of the CaCO(3) layer via pH-induced hydrolyzation allows glucose oxidase...	pmid:36055775 doi:10.1002/smll.202204244	Fri, 02 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
11	pubmed:36056015	SARS-CoV-2 mRNA-vaccine candidate; COReNAPCIN®, induces robust humoral and cellular immunity in mice and non-human primates	Reza Alimohammadi Meysam Porgoo Mohamad Eftekhary Seyed Hossein Kiaie Ehsan Ansari Dezfouli Maryam Dehghani Kaveh Nasrollahi Talieh Malekshahabi Maryam Heidari Sedigheh Pouya Masoumeh Alimohammadi Dorsa Sattari Khavas Mohammad Sadra Modaresi Mohammad Hossein Ghasemi Hamed Ramyar Fatemeh Mohammadipour Fateme Hamzelouei Ahmadreza Mofayezi Seyed Saeed Mottaghi Amirhosein Rahmati Mohsen Razzaznian Vista Tirandazi Mahdi Tat Fatemeh Borzouee Hossein Sadeghi Melika Haji Mohammadi Leila Rastegar Seyed Milad Safar Sajadi Hossein Ehsanbakhsh Hamed Bazmbar Zeinab Baghernejadan Maedeh Shams Nouraei Pouya Pazooki Mina Pahlavanneshan Khadijeh Alishah Fateme Nasiri Neda Mokhberian Seyedeh Shima Mohammadi Shima Akar Hamidreza Niknam Marzieh Azizi Mohammad Ajoudanian Mohammad Hossein Moteallehi-Ardakani Seyed Ali Mousavi Shaegh Reihaneh Ramezani Vahid Salimi Reza Moazzami Seyed Mahmoud Hashemi Somaye Dehghanizadeh Vahid Khoddami	At the forefront of biopharmaceutical industry, the messenger RNA (mRNA) technology offers a flexible and scalable platform to address the urgent need for world-wide immunization in pandemic situations. This strategic powerful platform has recently been used to immunize millions of people proving both of safety and highest level of clinical efficacy against infection with severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2). Here we provide preclinical report of COReNAPCIN^(®); a vaccine...	pmid:36056015 doi:10.1038/s41541-022-00528-3	Fri, 02 Sep 2022 06:00:00 -0400
12	pubmed:36056021	Transplantation of PSC-derived myogenic progenitors counteracts disease phenotypes in FSHD mice	Karim Azzag Darko Bosnakovski Sudheer Tungtur Peter Salama Michael Kyba Rita C R Perlingeiro	Facioscapulohumeral muscular dystrophy (FSHD) is a genetically dominant progressive myopathy caused by improper silencing of the DUX4 gene, leading to fibrosis, muscle atrophy, and fatty replacement. Approaches focused on muscle regeneration through the delivery of stem cells represent an attractive therapeutic option for muscular dystrophies. To investigate the potential for cell transplantation in FSHD, we have used the doxycycline-regulated iDUX4pA-HSA mouse model in which low-level DUX4 can...	pmid:36056021 doi:10.1038/s41536-022-00249-0	Fri, 02 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
13	pubmed:36056072	Xanthohumol exerts anti-inflammatory effects in an in vitro model of mechanically stimulated cementoblasts	Christian Niederau Shruti Bhargava Rebekka Schneider-Kramman Joachim Jankowski Rogerio B Craveiro Michael Wolf	Xanthohumol (XN) is a prenylated plant polyphenol that naturally occurs in hops and its products, e.g. beer. It has shown to have anti-inflammatory and angiogenesis inhibiting effects and it prevents the proliferation of cancer cells. These effects could be in particular interesting for processes within the periodontal ligament, as previous studies have shown that orthodontic tooth movement is associated with a sterile inflammatory reaction. Based on this, the study evaluates the...	pmid:36056072 doi:10.1038/s41598-022-19220-6	Fri, 02 Sep 2022 06:00:00 -0400
14	pubmed:36056084	Protein tyrosine kinase 2b inhibition reverts niche-associated resistance to tyrosine kinase inhibitors in AML	Catana Allert Alexander Waclawiczek Sarah Miriam Naomi Zimmermann Stefanie Göllner Daniel Heid Maike Janssen Simon Renders Christian Rohde Marcus Bauer Margarita Bruckmann Rafael Zinz Cornelius Pauli Birgit Besenbeck Claudia Wickenhauser Andreas Trumpp Jeroen Krijgsveld Carsten Müller-Tidow Maximilian Felix Blank	FLT3 tyrosine kinase inhibitor (TKI) therapy evolved into a standard therapy in FLT3-mutated AML. TKI resistance, however, develops frequently with poor outcomes. We analyzed acquired TKI resistance in AML cell lines by multilayered proteome analyses. Leupaxin (LPXN), a regulator of cell migration and adhesion, was induced during early resistance development, alongside the tyrosine kinase PTK2B which phosphorylated LPXN. Resistant cells differed in cell adhesion and migration, indicating altered...	pmid:36056084 doi:10.1038/s41375-022-01687-x	Fri, 02 Sep 2022 06:00:00 -0400
15	pubmed:36056180	Nervous system (NS) Tumors in Cancer Predisposition Syndromes	Prabhumallikarjun Patil Bojana Borislavova Pencheva Vinayak Mahesh Patil Jason Fangusaro	Genetic syndromes which develop one or more nervous system (NS) tumors as one of the manifestations can be grouped under the umbrella term of NS tumor predisposition syndromes. Understanding the underlying pathological pathways at the molecular level has led us to many radical discoveries, in understanding the mechanisms of tumorigenesis, tumor progression, interactions with the tumor microenvironment, and development of targeted therapies. Currently, at least 7-10% of all pediatric cancers are...	pmid:36056180 doi:10.1007/s13311-022-01277-w	Fri, 02 Sep 2022 06:00:00 -0400
16	pubmed:36056234	Dutch pharmacogenetics working group guideline for the gene-drug interaction of ABCG2, HLA-B and Allopurinol, and MTHFR, folic acid and methotrexate	Karel H van der Pol Marga Nijenhuis Bianca Soree Nienke J de Boer-Veger Anne Marie Buunk Henk-Jan Guchelaar Arne Risselada Ron H N van Schaik Jesse J Swen Daan Touw Jan van der Weide Roos van Westrhenen Vera H M Deneer Elisa J F Houwink Gerard A Rongen	The Dutch Pharmacogenetics Working Group (DPWG) aims to facilitate PGx implementation by developing evidence-based pharmacogenetics guidelines to optimize pharmacotherapy. This guideline describes the gene-drug interaction of ABCG2 with allopurinol, HLA-B with allopurinol, MTHFR with folic acid, and MTHFR with methotrexate, relevant for the treatment of gout, cancer, and rheumatoid arthritis. A systematic review was performed based on which pharmacotherapeutic recommendations were developed....	pmid:36056234 doi:10.1038/s41431-022-01180-0	Fri, 02 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
17	pubmed:36056297	STAT3 and PD-L1 are negatively correlated with ATM and have impact on the prognosis of triple-negative breast cancer patients with low ATM expression	Yuan-Ming Song Xiao-Long Qian Xiao-Qing Xia Ya-Qing Li Yuan-Yuan Sun Yu-Mian Jia Jin Wang Hui-Qin Xue Guang-Shen Gao Xiao-Zi Wang Xin-Min Zhang Xiao-Jing Guo	CONCLUSION: Locally advanced TNBC with low ATM expression may be more likely to benefit from anti-PD-L1 inhibitors. The feasibility of ATM functional inhibitor combined with immune checkpoint blockade therapies in the treatment of TNBC is also worthy of further exploration. Our study suggests that STAT3 has different impacts on tumor progression in different tumors.	pmid:36056297 doi:10.1007/s10549-022-06679-0	Fri, 02 Sep 2022 06:00:00 -0400
18	pubmed:36056383	Oct4 cooperates with c-Myc to improve mesenchymal-to-endothelial transition and myocardial repair of cardiac-resident mesenchymal stem cells	Lan Zhao Jianshuo Wang Pengzhen Wang Zhanyu Deng Jin Cui Weiguang Huang Shaoheng Zhang	CONCLUSIONS: Myocardial Isch drives resident cMSCs toward multiple phenotypes. Oct4 interacts with c-Myc to promote MEndoT capacity of cMSCs and improve their survival and reparative effects through upregulation of angiogenesis-related signaling pathways. These findings may identify novel targets for stem cell therapy.	pmid:36056383 doi:10.1186/s13287-022-03120-7	Fri, 02 Sep 2022 06:00:00 -0400
19	pubmed:36056395	Challenges and perspectives of tendon-derived cell therapy for tendinopathy: from bench to bedside	Ziming Chen Peilin Chen Monica Zheng Junjie Gao Delin Liu Allan Wang Qiujian Zheng Toby Leys Andrew Tai Minghao Zheng	Tendon is composed of dense fibrous connective tissues, connecting muscle at the myotendinous junction (MTJ) to bone at the enthesis and allowing mechanical force to transmit from muscle to bone. Tendon diseases occur at different zones of the tendon, including enthesis, MTJ and midsubstance of the tendon, due to a variety of environmental and genetic factors which consequently result in different frequencies and recovery rates. Self-healing properties of tendons are limited, and cell...	pmid:36056395 doi:10.1186/s13287-022-03113-6	Fri, 02 Sep 2022 06:00:00 -0400
20	pubmed:36056397	Biological characteristics and pulp regeneration potential of stem cells from canine deciduous teeth compared with those of permanent teeth	S M Ziauddin Misako Nakashima Hideto Watanabe Michiyo Tominaga Koichiro Iohara	CONCLUSIONS: These results demonstrated that DT-DPSCs could be a potential clinical alternative to PT-DPSCs for pulp regenerative therapy. DT-DPSCs can be preserved in an individual cell bank and used for potential future pulp regenerative therapy before the supply of an individual's own sound discarded teeth has been exhausted.	pmid:36056397 doi:10.1186/s13287-022-03124-3	Fri, 02 Sep 2022 06:00:00 -0400
21	pubmed:36056411	Natural history comparison study to assess the efficacy of elamipretide in patients with Barth syndrome	Brittany Hornby William Reid Thompson Mohammed Almuqbil Ryan Manuel Anthony Abbruscato Jim Carr Hilary J Vernon	CONCLUSIONS: Overall, the study established a NHC for use in assessing the efficacy of therapeutic interventions in patients with BTHS and the results suggest that elamipretide may improve natural history of BTHS at least in part by attenuating the natural decline in heart function and provide meaningful improvements in heart function and functional capacity in patients with BTHS compared to NHCs.	pmid:36056411 doi:10.1186/s13023-022-02469-5	Fri, 02 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
22	pubmed:36056635	Interaction between HER2 and ATM predicts poor survival in bladder cancer patients	Nada Albarakati Alaa Al-Shareeda Majed Ramadan Batla Al-Sowayan Ola Negm Taoufik Nedjadi	Human Epidermal Growth Factor Receptor 2 (HER2) overexpression is considered one of the interesting prognostic biomarkers in bladder cancer. However, the mechanism of bladder cancer development in relation to HER2 status remains to be elucidated. In this study, we investigated HER2-Ataxia telangiectasia mutated (ATM) kinase interaction and their impact on patient survival and cancer aggressiveness. Using the Cancer Genome Atlas (TCGA) cohorts, we demonstrated that ATM expression (protein/mRNA)...	pmid:36056635 doi:10.1111/jcmm.17512	Sat, 03 Sep 2022 06:00:00 -0400
23	pubmed:36056771	PBPK Model Development, Validation, and Application for Prediction of Eliglustat Drug-Drug Interactions	Siddhee A Sahasrabudhe Shen Cheng Mahmoud Al-Kofahi Jeanine R Jarnes Neal J Weinreb Reena V Kartha	Eliglustat is a glucosylceramide synthase inhibitor indicated as a long-term substrate reduction therapy for adults with type 1 Gaucher disease, a lysosomal rare disease. It is primarily metabolized by CYP2D6 and variants in the gene encoding this enzyme are important determinants of eliglustat pharmacokinetics (PK) and drug-drug interactions (DDIs). The existing drug label addresses the DDIs to some extent but has omitted scenarios where both metabolizing CYPs (2D6, 3A4) are mildly or...	pmid:36056771 doi:10.1002/cpt.2738	Sat, 03 Sep 2022 06:00:00 -0400
24	pubmed:36056785	Anti-proliferation effects of Apatinib in combination with Curcumin in breast cancer cells	Mahdi Farhoudi Sefidan Jadid Gholamreza Jahangirzadehd Javad Behroozi	CONCLUSIONS: In general, Apa-Cur combination therapy exerts more profound anti-proliferation effects on breast cancer cell than Apatinib or Curcumin monotherapy. However, further studies are required to identify other possible signaling pathways and mechanisms involved in the anticancer effects of Apatinib, Curcumin, and Apa-Cur.	pmid:36056785 doi:10.1515/hmbci-2022-0036	Sat, 03 Sep 2022 06:00:00 -0400
25	pubmed:36056851	CRISPR/Cas9-Based Gene Therapies for Fighting Drug Resistance Mediated by Cancer Stem Cellsc	Masoumeh Eliyasi Dashtaki Sorayya Ghasemi	Cancer stem cells (CSCs) are cancer-initiating cells found in most tumors and hematological cancers. CSCs are involved in cells progression, recurrence of tumors, and drug resistance. Current therapies have been focused on treating the mass of tumor cells and cannot eradicate the CSCs. CSCs drug-specific targeting is considered as an approach to precisely target these cells. Clustered regularly interspaced short palindromic repeats (CRISPR/Cas9) gene-editing systems are making progress and...	pmid:36056851 doi:10.2174/1566523222666220831161225	Sat, 03 Sep 2022 06:00:00 -0400
26	pubmed:36056858	Targeted treatment and immunotherapy in high-risk and relapsed/refractory pediatric acute lymphoblastic leukemia	Violeta Graiqevci-Uka Emir Behluli Lidvana Spahiu Thomas Liehr Gazmend Temaj	Acute lymphoblastic leukemia is the most frequent pediatric malignancy in children, comprising 30% of all pediatric malignancies; adult ALL comprises 5% of all ALL cases, which have a 186.6 per 1 million incidence. In pediatric ALL (pALL), on which this review focuses, approximately 1 in 285 children are diagnosed with cancer before the age of 20, and approximately 1 in 530 young adults between the ages of 20 and 39 years old is a childhood cancer survivor. The survival probability in pALL is...	pmid:36056858 doi:10.2174/1573396318666220901165247	Sat, 03 Sep 2022 06:00:00 -0400

	NCT Number	Title	Authors	Description	Identifier	Dates
27	pubmed:36056923	The mitochondrial seryl-tRNA synthetase SARS2 modifies onset in spastic paraplegia type 4	Livia Parodi Mathieu Barbier Maxime Jacoupy Claire Pujol François-Xavier Lejeune Pauline Lallemant-Dudek Typhaine Esteves Maartje Pennings Erik-Jan Kamsteeg Marine Guillaud-Bataille Guillaume Banneau Giulia Coarelli Badreddine Mohand Oumoussa Matthew J Fraidakis Giovanni Stevanin Christel Depienne Bart van de Warrenburg Alexis Brice Alexandra Durr	CONCLUSION: SARS2 overexpression lowers the age of onset in hereditary spastic paraplegia type 4. Lowering SARS2 or improving mitochondrial function could thus present viable approaches to therapy.	pmid:36056923 doi:10.1016/j.gim.2022.07.023	Sat, 03 Sep 2022 06:00:00 -0400
28	pubmed:36057072	Identifying potential causal effects of age at menopause: a Mendelian randomization phenome-wide association study	Maria C Magnus Maria Carolina Borges Abigail Fraser Deborah A Lawlor	Age at natural menopause (ANM) is associated with a range of health-related traits, including bone health, female reproductive cancers, and cardiometabolic health. Our objective was to conduct a Mendelian randomization phenome-wide association study (MR-pheWAS) of ANM. We conducted a hypothesis-free analysis of the genetic risk score (GRS) for ANM with 18,961 health-related traits among 181,279 women in UK Biobank. We also stratified the GRS according to the involvement of SNPs in DNA damage...	pmid:36057072 doi:10.1007/s10654-022-00903-3	Sat, 03 Sep 2022 06:00:00 -0400