## gene therapy

| | NCT Number | Title | Authors | Description | Identifier | Dates |
|---|---|---|---|---|---|---|
| 1 | pubmed:36049252 | Chondroitin sulfate modified chitosan nanoparticles as an efficient and targeted gene delivery vehicle to chondrocytes | Naghmeh Akbari Moghadam<br>Fatemeh Bagheri<br>Mohamadreza Baghaban Eslaminejad | Conventional treatments for osteoarthritis (OA), including drug delivery and tissue engineering approaches, could not offer a high yield of cartilage repair due to the compact and exclusive structure of cartilage. Targeted and high-efficiency delivery of gene sequences is necessary to rebalance the lost homeostatic properties of the cartilage in OA. Herein, we synthesized chitosan (CH)-chondroitin sulfate (CS) nanoparticles (NPs) as a platform for delivering gene sequences. These new... | pmid:36049252<br>doi:10.1016/j.colsurfb.2022.112786 | Thu, 01 Sep 2022 06:00:00 -0400 |
| 2 | pubmed:36049351 | Activation of AMP-activated protein kinase ablated the formation of aortic dissection by suppressing vascular inflammation and phenotypic switching of vascular smooth muscle cells | Lei Lei<br>Yanrong Zhou<br>Tiemao Wang<br>Zhi Zheng<br>Liang Chen<br>Youmin Pan | CONCLUSION: Impaired activation of AMPK may increase the susceptibility to aortic dissection. Our findings verified the protective effects of AMPK on the formation of aortic dissection and may provide evidence for clinical prevention or treatment. | pmid:36049351<br>doi:10.1016/j.intimp.2022.109177 | Thu, 01 Sep 2022 06:00:00 -0400 |
| 3 | pubmed:36049495 | Angiotensin receptor blockers and  blockers in Marfan syndrome: an individual patient data meta-analysis of randomised trials | Alex Pitcher<br>Enti Spata<br>Jonathan Emberson<br>Kelly Davies<br>Heather Halls<br>Lisa Holland<br>Kate Wilson<br>Christina Reith<br>Anne H Child<br>Tim Clayton<br>Matthew Dodd<br>Marcus Flather<br>Xu Yu Jin<br>George Sandor<br>Maarten Groenink<br>Barbara Mulder<br>Julie De Backer<br>Arturo Evangelista<br>Alberto Forteza<br>Gisela Teixido-Turà<br>Catherine Boileau<br>Guillaume Jondeau<br>Olivier Milleron<br>Ronald V Lacro<br>Lynn A Sleeper<br>Hsin-Hui Chiu<br>Mei-Hwan Wu<br>Stefan Neubauer<br>Hugh Watkins<br>Hal Dietz<br>Colin Baigent<br>Marfan Treatment Trialists’ Collaboration | BACKGROUND: Angiotensin receptor blockers (ARBs) and  blockers are widely used in the treatment of Marfan syndrome to try to reduce the rate of progressive aortic root enlargement characteristic of this condition, but their separate and joint effects are uncertain. We aimed to determine these effects in a collaborative individual patient data meta-analysis of randomised trials of these treatments. | pmid:36049495<br>doi:10.1016/S0140-6736(22)01534-3 | Thu, 01 Sep 2022 06:00:00 -0400 |
| 4 | pubmed:36049498 | Effect of statin therapy on muscle symptoms: an individual participant data meta-analysis of large-scale, randomised, double-blind trials | Cholesterol Treatment Trialists' Collaboration | BACKGROUND: Statin therapy is effective for the prevention of atherosclerotic cardiovascular disease and is widely prescribed, but there are persisting concerns that statin therapy might frequently cause muscle pain or weakness. We aimed to address these through an individual participant data meta-analysis of all recorded adverse muscle events in large, long-term, randomised, double-blind trials of statin therapy. | pmid:36049498<br>doi:10.1016/S0140-6736(22)01545-8 | Thu, 01 Sep 2022 06:00:00 -0400 |

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|---|---|---|---|---|---|---|
| 5 | pubmed:36049581 | Recent advances in HDAC-targeted imaging probes for cancer detection | Chu Tang<br>Xinan Wang<br>Yushen Jin<br>Fu Wang | Histone Deacetylases (HDACs) are abnormally high expressed in various cancers and play a crucial role in regulating gene expression. While HDAC-targeted inhibitors have been rapidly developed and approved in the last twenty years, noninvasive monitoring and visualizing the expression levels of HDACs in tumor tissues might help to early diagnosis in cancer and predict the response to HDAC-targeted cancer therapy. In this review, we summarize the recent advancements in the development of... | pmid:36049581<br>doi:10.1016/j.bbcan.2022.188788 | Thu, 01 Sep 2022 06:00:00 -0400 |
| 6 | pubmed:36049757 | Novel Therapeutic Agents for Rare Diseases of Calcium and Phosphate Metabolism | Afroditi Roumpou<br>Maria P Yavropoulou<br>Efstathios Chronopoulos<br>Eva Kassi | The last decade has been revolutionary regarding the management of rare bone diseases caused by impaired calcium and phosphate metabolism. Elucidation of the underlying genetic basis and pathophysiologic alterations has been the determinant factor for the development of new, disease-specific treatment agents. The phosphaturic hormone Fibroblast Growth Factor 23 (FGF23) possesses a critical role in the pathogenesis of various hypophosphatemic disorders. Among them, the genetic disorder of... | pmid:36049757<br>doi:10.1055/a-1917-0519 | Thu, 01 Sep 2022 06:00:00 -0400 |
| 7 | pubmed:36049896 | Expanded Clinical Pharmacogenetics Implementation Consortium (CPIC) Guideline for Medication Use in the Context of G6PD Genotype | Roseann S Gammal<br>Munir Pirmohamed<br>Andrew A Somogyi<br>Sarah A Morris<br>Christine M Formea<br>Amanda L Elchynski<br>Kazeem A Oshikoya<br>Howard L McLeod<br>Cyrine E Haidar<br>Michelle Whirl-Carrillo<br>Teri E Klein<br>Kelly E Caudle<br>Mary V Relling | Glucose-6-phosphate dehydrogenase (G6PD) deficiency is associated with development of acute hemolytic anemia in the setting of oxidative stress, which can be caused by medication exposure. Regulatory agencies worldwide warn against the use of certain medications in G6PD deficient persons, but in many cases, this information is conflicting, and the clinical evidence is sparse. This guideline provides information on using G6PD genotype as part of the diagnosis of G6PD deficiency and classifies... | pmid:36049896<br>doi:10.1002/cpt.2735 | Thu, 01 Sep 2022 06:00:00 -0400 |
| 8 | pubmed:36049972 | Paradigm shift in monogenic autoinflammatory diseases and systemic vasculitis: The VEXAS syndrome | José Hernández-Rodríguez<br>Anna Mensa-Vilaró<br>Juan I Aróstegui | VEXAS syndrome was described by the end of 2020 as an autoinflammatory disease caused by post-zygotic variants in the UBA1 gene. VEXAS syndrome occurs in adult males with recurrent fever, arthralgia/arthritis, ear/nose chondritis, neutrophilic dermatosis, lung inflammation, venous thrombosis, and different types of vasculitis. Common laboratory changes include raised acute phase reactants and macrocytic anemia. The coexistence of myelodysplasia is frequent, and bone marrow vacuolization of... | pmid:36049972<br>doi:10.1016/j.medcli.2022.06.018 | Thu, 01 Sep 2022 06:00:00 -0400 |

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| 9 | pubmed:36050142 | The evolving regulatory landscape in regenerative medicine | Danielle J Beetler<br>Damian N Di Florio<br>Ethan W Law<br>Chris M Groen<br>Anthony J Windebank<br>Quinn P Peterson<br>DeLisa Fairweather | Regenerative medicine as a field has emerged as a new component of modern medicine and medical research that encompasses a wide range of products including cellular and acellular therapies. As this new field emerged, regulatory agencies like the Food and Drug Administration (FDA) rapidly adapted existing regulatory frameworks to address the transplantation, gene therapy, cell-based therapeutics, and acellular biologics that fall under the broader regenerative medicine umbrella. Where it has not... | pmid:36050142<br>doi:10.1016/j.mam.2022.101138 | Thu, 01 Sep 2022 06:00:00 -0400 |
| 10 | pubmed:36050197 | Myeloid neoplasms associated with rearrangement of PDGFRB: A rare and tricky disease | Christophe Bontoux<br>Bouchra Badaoui<br>Nassera Abermil<br>Sihem Tarfi<br>Hélène Guermouche<br>Sydney Dubois<br>Lydia Roy<br>Juliette Vo Xuan<br>Violaine Tran Quang<br>Luojun Wang<br>Loetitia Favre<br>Elsa Poullot<br>Marc Michel<br>Ivan Sloma<br>Etienne Crickx<br>Adrien Pécriaux | In the latest World Health Organization classification (WHO), eosinophilic disorders represent a group of rare pathologic conditions with highly heterogeneous pathophysiology. In this report, we describe a case of myeloid neoplasm associated with eosinophilia and rearrangement of PDGFRB gene in a 67-year-old-male patient hospitalized with cerebellous ataxia. Initial investigations showed a bicytopenia with hypereosinophilia varying from 1.1 to 1.6×10/L. Bone marrow aspiration was rich and... | pmid:36050197<br>doi:10.1016/j.annpat.2022.03.005 | Thu, 01 Sep 2022 06:00:00 -0400 |
| 11 | pubmed:36050305 | Predicting remission after internet-delivered psychotherapy in patients with depression using machine learning and multi-modal data | John Wallert<br>Julia Boberg<br>Viktor Kaldo<br>David Mataix-Cols<br>Oskar Flygare<br>James J Crowley<br>Matthew Halvorsen<br>Fehmi Ben Abdesslem<br>Magnus Boman<br>Evelyn Andersson<br>Nils Hentati Isacsson<br>Ekaterina Ivanova<br>Christian Rück | This study applied supervised machine learning with multi-modal data to predict remission of major depressive disorder (MDD) after psychotherapy. Genotyped adult patients (n = 894, 65.5% women, age 18-75 years) diagnosed with mild-to-moderate MDD and treated with guided Internet-based Cognitive Behaviour Therapy (ICBT) at the Internet Psychiatry Clinic in Stockholm were included (2008-2016). Predictor types were demographic, clinical, process (e.g., time to complete online questionnaires), and... | pmid:36050305<br>doi:10.1038/s41398-022-02133-3 | Thu, 01 Sep 2022 06:00:00 -0400 |
| 12 | pubmed:36050451 | Adeno-associated virus mediated expression of monoclonal antibody MR191 protects mice against Marburg virus and provides long-term expression in sheep | Amira D Rghei<br>Laura P van Lieshout<br>Wenguang Cao<br>Shihua He<br>Kevin Tierney<br>Jordyn A Lopes<br>Nicole Zielinska<br>Enzo M Baracuhy<br>Elena S B Campbell<br>Jessica A Minott<br>Matthew M Guilleman<br>Pamela C Hasson<br>Brad Thompson<br>Khalil Karimi<br>Byram W Bridle<br>Leonardo Susta<br>Xiangguo Qiu<br>Logan Banadyga<br>Sarah K Wootton | Vectored monoclonal antibody (mAb) expression mediated by adeno-associated virus (AAV) gene delivery leads to sustained therapeutic mAb expression and protection against a wide range of infectious diseases in both small and large animal models, including nonhuman primates. Using our rationally engineered AAV6 triple mutant capsid, termed AAV6.2FF, we demonstrate rapid and robust expression of two potent human antibodies against Marburg virus, MR78 and MR191, following intramuscular (IM)... | pmid:36050451<br>doi:10.1038/s41434-022-00361-2 | Thu, 01 Sep 2022 06:00:00 -0400 |

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|---|---|---|---|---|---|---|
| 13 | pubmed:36050647 | Disease- and headache-specific microRNA signatures and their predicted mRNA targets in peripheral blood mononuclear cells in migraineurs: role of inflammatory signalling and oxidative stress | Timea Aczél<br>Bettina Benczik<br>Bence Ágg<br>Tamás Körtési<br>Péter Urbán<br>Witold Bauer<br>Attila Gyenesei<br>Bernadett Tuka<br>János Tajti<br>Péter Ferdinandy<br>László Vécsei<br>Kata Bölcskei<br>József Kun<br>Zsuzsanna Helyes | CONCLUSIONS: We provide here the first evidence for disease- and headache-specific miRNA signatures in the PBMC of migraineurs, which might help to identify novel targets for both prophylaxis and attack therapy. | pmid:36050647<br>doi:10.1186/s10194-022-01478-w | Thu, 01 Sep 2022 06:00:00 -0400 |
| 14 | pubmed:36050710 | Developing machine learning-based models to predict intrauterine insemination (IUI) success by address modeling challenges in imbalanced data and providing modification solutions for them | Sajad Khodabandelu<br>Zahra Basirat<br>Sara Khaleghi<br>Soraya Khafri<br>Hussain Montazery Kordy<br>Masoumeh Golsorkhtabaramiri | CONCLUSION: The results of this study with the XGBoost prediction model can be used to foretell the individual success of IUI for each couple before initiating therapy. | pmid:36050710<br>doi:10.1186/s12911-022-01974-8 | Thu, 01 Sep 2022 06:00:00 -0400 |
| 15 | pubmed:36050749 | Delayed diagnosis of complex glycerol kinase deficiency in a Chinese male infant: a case report | Na Tao<br>Xiaomei Liu<br>Yueqi Chen<br>Meiyuan Sun<br>Fang Xu<br>Yanfang Su | CONCLUSIONS: Overall, CGKD, although rare, cannot be easily excluded in children with persistent vomiting. Extensive blood tests can help to detect abnormal indicators. Adrenal crisis needs to be avoided as much as possible during corticosteroid replacement therapy. | pmid:36050749<br>doi:10.1186/s12887-022-03568-9 | Thu, 01 Sep 2022 06:00:00 -0400 |
| 16 | pubmed:36050760 | Impaired mitochondria of Tregs decreases OXPHOS-derived ATP in primary immune thrombocytopenia with positive plasma pathogens detected by metagenomic sequencing | Yanxia Zhan<br>Jingjing Cao<br>Lili Ji<br>Miaomiao Zhang<br>Qi Shen<br>Pengcheng Xu<br>Xibing Zhuang<br>Shanshan Qin<br>Fanli Hua<br>Lihua Sun<br>Feng Li<br>Hao Chen<br>Yunfeng Cheng | CONCLUSIONS: Impaired mitochondria function of Tregs in positive pathogen-ITP patients caused a decrease of OXPHOS-derived ATP and overall metabolism flux that might be the cause of steroid resistance in ITP patients. | pmid:36050760<br>doi:10.1186/s40164-022-00304-y | Thu, 01 Sep 2022 06:00:00 -0400 |
| 17 | pubmed:36051022 | Hematopoietic stem progenitor cells with malignancy-related gene mutations in patients with acquired aplastic anemia are characterized by the increased expression of CXCR4 | Takamasa Katagiri<br>Jorge Luis Espinoza<br>Mizuho Uemori<br>Honoka Ikeda<br>Kohei Hosokawa<br>Ken Ishiyama<br>Takeshi Yoroidaka<br>Tatsuya Imi<br>Hiroyuki Takamatsu<br>Tatsuhiko Ozawa<br>Hiroyuki Kishi<br>Yasuhiko Yamamoto<br>Mahmoud Ibrahim Elbadry<br>Yoshinori Yoshida<br>Kazuhisa Chonabayashi<br>Katsuto Takenaka<br>Koichi Akashi<br>Yasuhito Nannya<br>Seishi Ogawa<br>Shinji Nakao | The phenotypic changes in hematopoietic stem progenitor cells (HSPCs) with somatic mutations of malignancy-related genes in patients with acquired aplastic anemia (AA) are poorly understood. As our initial study showed increased CXCR4 expression on HLA allele-lacking (HLA[-]) HSPCs that solely support hematopoiesis in comparison to redundant HLA(+) HSPCs in AA patients, we screened the HSPCs of patients with various types of bone marrow (BM) failure to investigate their CXCR4 expression. In... | pmid:36051022<br>pmc:PMC9422028<br>doi:10.1002/jha2.515 | Fri, 02 Sep 2022 06:00:00 -0400 |

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| 18 | pubmed:36051037 | B-cell receptors of EBV-negative Burkitt lymphoma bind modified isoforms of autoantigens | Theresa Bock<br>Moritz Bewarder<br>Onur Cetin<br>Natalie Fadle<br>Evi Regitz<br>Eva C Schwarz<br>Jana Held<br>Sophie Roth<br>Stefan Lohse<br>Thorsten Pfuhl<br>Rabea Wagener<br>Sigrun Smola<br>Sören L Becker<br>Rainer Maria Bohle<br>Lorenz Trümper<br>Reiner Siebert<br>Martin-Leo Hansmann<br>Michael Pfreundschuh<br>Hans G Drexler<br>Markus Hoth<br>Boris Kubuschok<br>Klaus Roemer<br>Klaus-Dieter Preuss<br>Sylvia Hartmann<br>Lorenz Thurner | Burkitt lymphoma (BL) represents the most aggressive B-cell-lymphoma. Beside the hallmark of IG-MYC-translocation, surface B-cell receptor (BCR) is expressed, and mutations in the BCR pathway are frequent. Coincidental infections in endemic BL, and specific extra-nodal sites suggest antigenic triggers. To explore this hypothesis, BCRs of BL cell lines and cases were screened for reactivities against a panel of bacterial lysates, lysates of Plasmodium falciparum, a custom-made virome array and... | pmid:36051037<br>pmc:PMC9421956<br>doi:10.1002/jha2.475 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 19 | pubmed:36051053 | microRNA expression in acute myeloid leukaemia: New targets for therapy? | Daniel Fletcher<br>Elliott Brown<br>Julliah Javadala<br>Pinar Uysal-Onganer<br>Barbara-Ann Guinn | Recent studies have shown that short non-coding RNAs, known as microRNAs (miRNAs) and their dysregulation, are implicated in the pathogenesis of acute myeloid leukaemia (AML). This is due to their role in the control of gene expression in a variety of molecular pathways. Therapies involving miRNA suppression and replacement have been developed. The normalisation of expression and the subsequent impact on AML cells have been investigated for some miRNAs, demonstrating their potential to act as... | pmid:36051053<br>pmc:PMC9421970<br>doi:10.1002/jha2.441 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 20 | pubmed:36051057 | The 17-gene stemness score associates with relapse risk and long-term outcomes following allogeneic haematopoietic cell transplantation in acute myeloid leukaemia | Dennis D H Kim<br>Igor Novitzky Basso<br>Taehyung Simon Kim<br>Seong Yoon Yi<br>Kyoung Ha Kim<br>Tracy Murphy<br>Steven Chan<br>Mark Minden<br>Ivan Pasic<br>Wilson Lam<br>Arjun Law<br>Fotios V Michelis<br>Armin Gerbitz<br>Auro Viswabandya<br>Jeffrey Lipton<br>Rajat Kumar<br>Stanley W K Ng<br>Tracy Stockley<br>Tong Zhang<br>Ian King<br>Jonas Mattsson<br>Jean C Y Wang | A 17-gene stemness (LSC17) score determines risk in acute myeloid leukaemia patients treated with standard chemotherapy regimens. The present study further analysed the impact of the LSC17 score at diagnosis on outcomes following allogeneic haematopoietic cell transplantation (HCT). Out of 452 patients with available LSC17 score, 123 patients received allogeneic HCT. Transplant outcomes, including overall (OS), leukaemia-free survival (LFS), relapse incidence (RI) and non-relapse mortality... | pmid:36051057<br>pmc:PMC9422016<br>doi:10.1002/jha2.466 | Fri, 02 Sep 2022 06:00:00 -0400 |

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| 21 | pubmed:36051067 | Transcriptional profiles define drug refractory disease in myeloma | Yuan Xiao Zhu<br>Laura A Bruins<br>Xianfeng Chen<br>Chang-Xin Shi<br>Cecilia Bonolo De Campos<br>Nathalie Meurice<br>Xuewei Wang<br>Greg J Ahmann<br>Colleen A Ramsower<br>Esteban Braggio<br>Lisa M Rimsza<br>A Keith Stewart | Identifying biomarkers associated with disease progression and drug resistance are important for personalized care. We investigated the expression of 121 curated genes, related to immunomodulatory drugs (IMiDs) and proteasome inhibitors (PIs) responsiveness. We analyzed 28 human multiple myeloma (MM) cell lines with known drug sensitivities and 130 primary MM patient samples collected at different disease stages, including newly diagnosed (ND), on therapy (OT), and relapsed and refractory (RR,... | pmid:36051067<br>pmc:PMC9422020<br>doi:10.1002/jha2.455 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 22 | pubmed:36051087 | Heterogeneous genetic and non-genetic mechanisms contribute to response and resistance to azacitidine monotherapy | Vasiliki Symeonidou<br>Marlen Metzner<br>Batchimeg Usukhbayar<br>Aimee E Jackson<br>Sonia Fox<br>Charles F Craddock<br>Paresh Vyas | Acute myeloid leukaemia is prevalent in older patients that are often ineligible for intensive chemotherapy and treatment options remain limited with azacitidine being at the forefront. Azacitidine has been used in the clinic for decades, however, we still lack a complete understanding of the mechanisms by which the drug exerts its anti-tumour effect. To gain insight into the mechanism of action, we defined the mutational profile of sequential samples of patients treated with azacitidine. We did... | pmid:36051087<br>pmc:PMC9421974<br>doi:10.1002/jha2.527 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 23 | pubmed:36051114 | Network pharmacology-based strategy for predicting therapy targets of Sanqi and Huangjing in diabetes mellitus | Xiao-Yan Cui<br>Xiao Wu<br>Dan Lu<br>Dan Wang | CONCLUSION: SQHJ treatment for DM targets TP53, AKT1, CASP3, and TNF and participates in pathways in leishmaniasis and cancer. | pmid:36051114<br>pmc:PMC9297423<br>doi:10.12998/wjcc.v10.i20.6900 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 24 | pubmed:36051141 | Novel mutation in the *SALL1* gene in a four-generation Chinese family with uraemia: A case report | Jia-Xi Fang<br>Jin-Shi Zhang<br>Min-Min Wang<br>Lin Liu | CONCLUSION: We report a novel SALL1 exon 2 (c.3437delG) mutation and clinical syndrome with kidney disease, bilateral overlapping toes, unilateral dysplastic external ears, and sensorineural hearing loss in a four-generation Chinese family. | pmid:36051141<br>pmc:PMC9297417<br>doi:10.12998/wjcc.v10.i20.7068 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 25 | pubmed:36051248 | Rescue of the Congenital Hereditary Endothelial Dystrophy Mouse Model by Adeno-Associated Viruse-Mediated *Slc4a11* Replacement | Rajalekshmy Shyam<br>Diego G Ogando<br>Edward T Kim<br>Subashree Murugan<br>Moonjung Choi<br>Joseph A Bonanno | CONCLUSIONS: Functional rescue of CHED phenotypes in the Slc4a11 ^(-/-) mouse is possible; however, early intervention is critical. | pmid:36051248<br>pmc:PMC9432820<br>doi:10.1016/j.xops.2021.100084 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 26 | pubmed:36051285 | Case report: Transplantation of human induced pluripotent stem cell-derived cardiomyocyte patches for ischemic cardiomyopathy | Shigeru Miyagawa<br>Satoshi Kainuma<br>Takuji Kawamura<br>Kota Suzuki<br>Yoshito Ito<br>Hiroko Iseoka<br>Emiko Ito<br>Maki Takeda<br>Masao Sasai<br>Noriko Mochizuki-Oda<br>Tomomi Shimamoto<br>Yukako Nitta<br>Hiromi Dohi<br>Tadashi Watabe<br>Yasushi Sakata<br>Koichi Toda<br>Yoshiki Sawa | Despite major therapeutic advances, heart failure, as a non-communicable disease, remains a life-threatening disorder, with 26 million patients worldwide, causing more deaths than cancer. Therefore, novel strategies for the treatment of heart failure continue to be an important clinical need. Based on preclinical studies, allogenic human induced pluripotent stem cell-derived cardiomyocyte (hiPSC-CM) patches have been proposed as a potential therapeutic candidate for heart failure. We report the... | pmid:36051285<br>pmc:PMC9426776<br>doi:10.3389/fcvm.2022.950829 | Fri, 02 Sep 2022 06:00:00 -0400 |

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| 27 | pubmed:36051286 | Pediatric patients with familially inherited sitosterolemia: Two case reports | Shun-Qing Su<br>Di-Sheng Xiong<br>Xiu-Mei Ding<br>Jin-An Kuang<br>Yue-Chun Lin | CONCLUSION: Sitosterolemia is an autosomal recessive disease that could be effectively controlled after dietary control and oral lipid-lowering therapy with Ezetimibe. Xanthomas, which affects function and appearance, could be surgically removed, and primary wound healing could be achieved. | pmid:36051286<br>pmc:PMC9424688<br>doi:10.3389/fcvm.2022.927267 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 28 | pubmed:36051386 | Primary aldosteronism caused by a pI157S somatic *KCNJ5* mutation in a black adolescent female with aldosterone-producing adenoma | Celso E Gomez-Sanchez<br>Desmaré van Rooyen<br>William E Rainey<br>Kazutaka Nanba<br>Amy R Blinder<br>Radhakrishna Baliga | Aldosterone-producing adenoma is a rare cause of hypertension in children. Only a limited number of cases of aldosterone-producing adenomas with somatic KCNJ5 gene mutations have been described in children. Blacks are particularly more susceptible to developing long-standing cardiovascular effects of aldosterone-induced severe hypertension. Somatic CACNA1D gene mutations are particularly more prevalent in black males whereas KCNJ5 gene mutations are most frequently present in black females. We... | pmid:36051386<br>pmc:PMC9424617<br>doi:10.3389/fendo.2022.921449 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 29 | pubmed:36051387 | Effects of *PPARD* gene variants on the therapeutic responses to exenatide in chinese patients with type 2 diabetes mellitus | Jinfang Song<br>Na Li<br>Ruonan Hu<br>Yanan Yu<br>Ke Xu<br>Hongwei Ling<br>Qian Lu<br>Tingting Yang<br>Tao Wang<br>Xiaoxing Yin | CONCLUSION: These data suggest that the PPARD rs2016520 and rs3777744 polymorphisms are associated with exenatide monotherapy efficacy, due to the pivotal role of PPAR in regulating insulin resistance through affecting the expression of GLP-1R. This study was registered in the Chinese Clinical Trial Register (No. ChiCTR-CCC13003536). | pmid:36051387<br>pmc:PMC9424689<br>doi:10.3389/fendo.2022.949990 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 30 | pubmed:36051396 | Dental health of pediatric patients with X-linked hypophosphatemia (XLH) after three years of burosumab therapy | Rafi Brener<br>Leonid Zeitlin<br>Yael Lebenthal<br>Avivit Brener | An inactivating PHEX gene mutation with the resultant accumulation of several mineralization-inhibiting proteins (e.g., FGF23) causes skeletal and dental morbidity in X-linked hypophosphatemia (XLH). This prospective case-control study explored the effect of burosumab, an anti-FGF23 antibody, on dental health of children with XLH. Ten children (age 4.3-15 years) with XLH underwent burosumab treatment per protocol. Assessment of their dental status at treatment initiation and after 1 and 3 years... | pmid:36051396<br>pmc:PMC9425915<br>doi:10.3389/fendo.2022.947814 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 31 | pubmed:36051466 | ENTPD1/CD39 as a predictive marker of treatment response to gemogenovatucel-T as maintenance therapy in newly diagnosed ovarian cancer | Rodney P Rocconi<br>Laura Stanbery<br>Min Tang<br>Adam Walter<br>Bradley J Monk<br>Thomas J Herzog<br>Robert L Coleman<br>Luisa Manning<br>Gladice Wallraven<br>Staci Horvath<br>Ernest Bognar<br>Neil Senzer<br>Scott Brun<br>John Nemunaitis | CONCLUSION: NSA should be considered for application to investigational targeted therapies in order to identify populations most likely to benefit from treatment, in preparation for efficacy conclusive trials. | pmid:36051466<br>pmc:PMC9424215<br>doi:10.1038/s43856-022-00163-y | Fri, 02 Sep 2022 06:00:00 -0400 |

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| 32 | pubmed:36051614 | PARP inhibitors on the move in prostate cancer: spotlight on Niraparib & update on PARP inhibitor combination trials | Erica L Beatson<br>Cindy H Chau<br>Douglas K Price<br>William D Figg | PARP inhibitors were recently introduced as a novel targeted therapy for biomarker positive metastatic castration resistant prostate cancer (mCRPC) patients, a population that inevitably acquires resistance to existing standard care regimens. Olaparib and rucaparib are now FDA-approved for mCRPC, while talazoparib and niraparib are advancing through the clinical stage of development. We highlight the recent results of the GALAHAD trial testing the efficacy of niraparib in mCRPC patients with DNA... | pmid:36051614<br>pmc:PMC9428566 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 33 | pubmed:36051659 | Endoplasmic-reticulum-stress-induced lipotoxicity in human kidney epithelial cells | Tuçe Çeker<br>Çaatay Ylmaz<br>Esma Krmloglu<br>Mutay Aslan | Accumulation of lipids and their intermediary metabolites under endoplasmic reticulum (ER) stress instigates metabolic failure, described as lipotoxicity, in the kidney. This study aimed to determine ER-stress-related sphingolipid and polyunsaturated fatty acid (PUFA) changes in human kidney cells. Tunicamycin (TM) was employed to induce ER stress and an ER stress inhibitor, tauroursodeoxycholic acid (TUDCA), was given to minimize cytotoxicity. Cell viability was determined by MTT assay.... | pmid:36051659<br>pmc:PMC9424710<br>doi:10.1093/toxres/tfac041 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 34 | pubmed:36051694 | Computational Analysis of Deleterious SNPs in *NRAS* to Assess Their Potential Correlation With Carcinogenesis | Mohammed Y Behairy<br>Mohamed A Soltan<br>Mohamed S Adam<br>Ahmed M Refaat<br>Ehab M Ezz<br>Sarah Albogami<br>Eman Fayad<br>Fayez Althobaiti<br>Ahmed M Gouda<br>Ashraf E Sileem<br>Mahmoud A Elfaky<br>Khaled M Darwish<br>Muhammad Alaa Eldeen | The NRAS gene is a well-known oncogene that acts as a major player in carcinogenesis. Mutations in the NRAS gene have been linked to multiple types of human tumors. Therefore, the identification of the most deleterious single nucleotide polymorphisms (SNPs) in the NRAS gene is necessary to understand the key factors of tumor pathogenesis and therapy. We aimed to retrieve NRAS missense SNPs and analyze them comprehensively using sequence and structure approaches to determine the most deleterious... | pmid:36051694<br>pmc:PMC9424727<br>doi:10.3389/fgene.2022.872845 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 35 | pubmed:36051701 | A homozygous variant in the *GPIHBP1* gene in a child with severe hypertriglyceridemia and a systematic literature review | Ursa Sustar<br>Urh Groselj<br>Sabeen Abid Khan<br>Saeed Shafi<br>Iqbal Khan<br>Jernej Kovac<br>Barbara Jenko Bizjan<br>Tadej Battelino<br>Fouzia Sadiq | Background: Due to nonspecific symptoms, rare dyslipidaemias are frequently misdiagnosed, overlooked, and undertreated, leading to increased risk for severe cardiovascular disease, pancreatitis and/or multiple organ failures before diagnosis. Better guidelines for the recognition and early diagnosis of rare dyslipidaemias are urgently required. Methods: Genomic DNA was isolated from blood samples of a Pakistani paediatric patient with hypertriglyceridemia, and from his parents and siblings.... | pmid:36051701<br>pmc:PMC9424485<br>doi:10.3389/fgene.2022.983283 | Fri, 02 Sep 2022 06:00:00 -0400 |

| | NCT Number | Title | Authors | Description | Identifier | Dates |
|---|---|---|---|---|---|---|
| 36 | pubmed:36051751 | CDK4: a master regulator of the cell cycle and its role in cancer | Stacey J Baker<br>Poulikos I Poulikakos<br>Hanna Y Irie<br>Samir Parekh<br>E Premkumar Reddy | The cell cycle is regulated in part by cyclins and their associated serine/threonine cyclin-dependent kinases, or CDKs. CDK4, in conjunction with the D-type cyclins, mediates progression through the G(1) phase when the cell prepares to initiate DNA synthesis. Although Cdk4-null mutant mice are viable and cell proliferation is not significantly affected in vitro due to compensatory roles played by other CDKs, this gene plays a key role in mammalian development and cancer. This review discusses... | pmid:36051751<br>pmc:PMC9426627<br>doi:10.18632/genesandcancer.221 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 37 | pubmed:36051855 | Nanotechnology Advances in the Detection and Treatment of Cancer: An Overview | Sareh Mosleh-Shirazi<br>Milad Abbasi<br>Mohammad Reza Moaddeli<br>Ahmad Vaez<br>Mostafa Shafiee<br>Seyed Reza Kasaee<br>Ali Mohammad Amani<br>Saeid Hatam | Over the last few years, progress has been made across the nanomedicine landscape, in particular, the invention of contemporary nanostructures for cancer diagnosis and overcoming complexities in the clinical treatment of cancerous tissues. Thanks to their small diameter and large surface-to-volume proportions, nanomaterials have special physicochemical properties that empower them to bind, absorb and transport high-efficiency substances, such as small molecular drugs, DNA, proteins, RNAs, and... | pmid:36051855<br>pmc:PMC9428923<br>doi:10.7150/ntno.74613 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 38 | pubmed:36051870 | MLACP 2.0: An updated machine learning tool for anticancer peptide prediction | Le Thi Phan<br>Hyun Woo Park<br>Thejkiran Pitti<br>Thirumurthy Madhavan<br>Young-Jun Jeon<br>Balachandran Manavalan | Anticancer peptides are emerging anticancer drug that offers fewer side effects and is more effective than chemotherapy and targeted therapy. Predicting anticancer peptides from sequence information is one of the most challenging tasks in immunoinformatics. In the past ten years, machine learning-based approaches have been proposed for identifying ACP activity from peptide sequences. These methods include our previous method MLACP (developed in 2017) which made a significant impact on anticancer... | pmid:36051870<br>pmc:PMC9421197<br>doi:10.1016/j.csbj.2022.07.043 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 39 | pubmed:36051887 | Network metrics, structural dynamics and density functional theory calculations identified a novel Ursodeoxycholic Acid derivative against therapeutic target Parkin for Parkinson's disease | Aniket Naha<br>Sanjukta Banerjee<br>Reetika Debroy<br>Soumya Basu<br>Gayathri Ashok<br>P Priyamvada<br>Hithesh Kumar<br>A R Preethi<br>Harpreet Singh<br>Anand Anbarasu<br>Sudha Ramaiah | Parkinson's disease (PD) has been designated as one of the priority neurodegenerative disorders worldwide. Although diagnostic biomarkers have been identified, early onset detection and targeted therapy are still limited. An integrated systems and structural biology approach were adopted to identify therapeutic targets for PD. From a set of 49 PD associated genes, a densely connected interactome was constructed. Based on centrality indices, degree of interaction and functional enrichments,... | pmid:36051887<br>pmc:PMC9399899<br>doi:10.1016/j.csbj.2022.08.017 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 40 | pubmed:36051970 | Longitudinal neurological analysis of moderate and severe pediatric cerebral visual impairment | Andres Jimenez-Gomez<br>Kristen S Fisher<br>Kevin X Zhang<br>Chunyan Liu<br>Qin Sun<br>Veeral S Shah | CONCLUSION: This study offers extensive insights into neurologic, developmental and ophthalmologic features in patients with moderate to severe CVI. In concordance with previous findings, aspects of perinatal history and epilepsy/seizure control may help inform severity and prognosis in the general neurology or ophthalmology clinic. Conversely, these aspects, as well as genetic and specific epilepsy traits may alert vision health care providers in the clinic to pursue visual evaluation in... | pmid:36051970<br>pmc:PMC9425457<br>doi:10.3389/fnhum.2022.772353 | Fri, 02 Sep 2022 06:00:00 -0400 |

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|---|---|---|---|---|---|---|
| 41 | pubmed:36052090 | SIRGs score may be a predictor of prognosis and immunotherapy response for esophagogastric junction adenocarcinoma | Li-Ying OuYang<br>Zi-Jian Deng<br>Yu-Feng You<br>Jia-Ming Fang<br>Xi-Jie Chen<br>Jun-Jie Liu<br>Xian-Zhe Li<br>Lei Lian<br>Shi Chen | CONCLUSION: The SIRGs score may be a predictor of the prognosis and immune-therapy response for esophagogastric junction adenocarcinoma. | pmid:36052090<br>pmc:PMC9424497<br>doi:10.3389/fimmu.2022.977894 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 42 | pubmed:36052091 | Targeted treatment of autoimmune cytopenias in primary immunodeficiencies | Lucia Pacillo<br>Giuliana Giardino<br>Donato Amodio<br>Carmela Giancotta<br>Beatrice Rivalta<br>Gioacchino Andrea Rotulo<br>Emma Concetta Manno<br>Cristina Cifaldi<br>Giuseppe Palumbo<br>Claudio Pignata<br>Paolo Palma<br>Paolo Rossi<br>Andrea Finocchi<br>Caterina Cancrini | Primary Immunodeficiencies (PID) are a group of rare congenital disorders of the immune system. Autoimmune cytopenia (AIC) represents the most common autoimmune manifestation in PID patients. Treatment of AIC in PID patients can be really challenging, since they are often chronic, relapsing and refractory to first line therapies, thus requiring a broad variety of alternative therapeutic options. Moreover, immunosuppression should be fine balanced considering the increased susceptibility to... | pmid:36052091<br>pmc:PMC9426461<br>doi:10.3389/fimmu.2022.911385 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 43 | pubmed:36052149 | Comparison of Gene Editing versus a Neutrophil Elastase Inhibitor as Potential Therapies for *ELANE* Neutropenia | Vahagn Makaryan<br>Merideth Kelley<br>Breanna Fletcher<br>Isabella Archibald<br>Tanoya Poulsen<br>David Dale | Heterozygous mutations in ELANE, the gene for neutrophil elastase, cause cyclic and congenital neutropenia through the programed cell death of neutrophil progenitors in the bone marrow. Granulocyte colony-stimulating factor is an effective therapy for these diseases, but alternative therapies are needed, especially for patients who do not respond well or are at high risk of developing myeloid malignancies. We developed an HL60 cell model for ELANE neutropenia and previously demonstrated that... | pmid:36052149<br>pmc:PMC9431957<br>doi:10.33696/immunology.4.129 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 44 | pubmed:36052158 | Identification of a Prognostic Model Based on Immune Cell Signatures in Clear Cell Renal Cell Carcinoma | Xuezhong Shi<br>Yali Niu<br>Yongli Yang<br>Nana Wang<br>Mengyang Yuan<br>Chaojun Yang<br>Ani Dong<br>Huili Zhu<br>Xiaocan Jia | CONCLUSION: The ICS score model has higher predictive power for patients' prognosis and can instruct ccRCC patients in seeking suitable treatment. | pmid:36052158<br>pmc:PMC9427244<br>doi:10.1155/2022/1727575 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 45 | pubmed:36052243 | Autophagy-related prognostic signature characterizes tumor microenvironment and predicts response to ferroptosis in gastric cancer | Haoran Li<br>Bing Xu<br>Jing Du<br>Yunyi Wu<br>Fangchun Shao<br>Yan Gao<br>Ping Zhang<br>Junyu Zhou<br>Xiangmin Tong<br>Ying Wang<br>Yanchun Li | CONCLUSIONS: Autophagy patterns are involved in TME diversity and complexity. Autophagy score can be used as an independent prognostic biomarker in GC patients and to predict the effect of immunotherapy and ferroptosis-based therapy. This might benefit individualized treatment for GC. | pmid:36052243<br>pmc:PMC9424910<br>doi:10.3389/fonc.2022.959337 | Fri, 02 Sep 2022 06:00:00 -0400 |

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|---|---|---|---|---|---|---|
| 46 | pubmed:36052251 | Minimal residual disease detection by next-generation sequencing in multiple myeloma: Promise and challenges for response-adapted therapy | Valeria Ferla<br>Elena Antonini<br>Tommaso Perini<br>Francesca Farina<br>Serena Masottini<br>Simona Malato<br>Sarah Marktel<br>Maria Teresa Lupo Stanghellini<br>Cristina Tresoldi<br>Fabio Ciceri<br>Magda Marcatti | Assessment of minimal residual disease (MRD) is becoming a standard diagnostic tool for curable hematological malignancies such as chronic and acute myeloid leukemia. Multiple myeloma (MM) remains an incurable disease, as a major portion of patients even in complete response eventually relapse, suggesting that residual disease remains. Over the past decade, the treatment landscape of MM has radically changed with the introduction of new effective drugs and the availability of immunotherapy,... | pmid:36052251<br>pmc:PMC9426755<br>doi:10.3389/fonc.2022.932852 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 47 | pubmed:36052254 | Lentiviral vector-based xenograft tumors as candidate reference materials for detection of HER2-low breast cancer | Yali Wei<br>Xu An<br>Qinmei Cao<br>Nanying Che<br>Yuanyuan Xue<br>Haiteng Deng<br>Qingtao Wang<br>Rui Zhou | The human epidermal growth factor receptor 2 (HER2) is an important biomarker that plays a pivotal role in therapeutic decision-making for patients with breast cancer (BC). Patients with HER2-low BC can benefit from new HER2 targeted therapy. For ensuring the accurate and reproducible detection of HER2-low cancer, reliable reference materials are required for monitoring the sensitivity and specificity of detection assays. Herein, a lentiviral vector was used to transduce the HER2 gene into... | pmid:36052254<br>pmc:PMC9425432<br>doi:10.3389/fonc.2022.955943 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 48 | pubmed:36052274 | Study on Biomarkers Related to the Treatment of Post-Stroke Depression and Alternative Medical Treatment Methods | Menghan Li<br>Ran Ding<br>Xinming Yang<br>Dawei Ran | CONCLUSION: The results of this study show that five genes ("NRBP1", "SIRT1", "BDNF", "MAPK3", "CREB1".) and key biological pathways such as NFkB, PI3K/AKT activation, and MAPK are the keys to the occurrence and development of PSD biomarkers, which can also be therapeutically intervened by acupuncture. | pmid:36052274<br>pmc:PMC9426768<br>doi:10.2147/NDT.S370848 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 49 | pubmed:36052279 | Identification of a Hypoxia-Angiogenesis lncRNA Signature Participating in Immunosuppression in Gastric Cancer | Zicheng Wang<br>Xisong Liang<br>Hao Zhang<br>Zeyu Wang<br>Xun Zhang<br>Ziyu Dai<br>Zaoqu Liu<br>Jian Zhang<br>Peng Luo<br>Jiarong Li<br>Quan Cheng | Hypoxia and angiogenesis are the leading causes of tumor progression, and their strong correlation has been discovered in many cancers. However, their collective function's prognostic and biological roles were not reported in gastric cancer. Hence, we aimed to investigate the effects of hypoxia and angiogenesis on gastric cancer via sequencing data. This study used weighted gene coexpression network analysis and random forest regression to build a hypoxia-angiogenesis-related model (HARM) via... | pmid:36052279<br>pmc:PMC9427269<br>doi:10.1155/2022/5209607 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 50 | pubmed:36052356 | Treatment of latent tuberculosis in a child with mucopolysaccharidosis type I receiving enzyme replacement therapy: A case report | Lauma Vasilevska<br>Madara Kreile<br>Ieva Grinfelde<br>Anita Skangale | Mucopolysaccharidosis type I S (MPS IS) is a rare autosomal recessive lysosomal storage disorder caused by mutations in the IDUA gene, leading to a deficiency of the enzyme alpha-L-iduronidase. Enzyme replacement therapy (ERT) reduces lysosomal storage in the liver and improves clinical manifestations. To date, there are no published reports of tuberculosis (TB) treatment in MPS IS patients receiving ERT and as such it is not known whether both conditions can be treated simultaneously. Here, we... | pmid:36052356<br>pmc:PMC9424730<br>doi:10.3389/fped.2022.973193 | Fri, 02 Sep 2022 06:00:00 -0400 |

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|---|---|---|---|---|---|---|
| 51 | pubmed:36052448 | Expanded clinical spectrum of oculopharyngodistal myopathy type 1 | Takahiro Shimizu<br>Hiroyuki Ishiura<br>Manato Hara<br>Shota Shibata<br>Atsushi Unuma<br>Akatsuki Kubota<br>Kaori Sakuishi<br>Kiyoharu Inoue<br>Jun Goto<br>Yuji Takahashi<br>Yuichiro Shirota<br>Masashi Hamada<br>Jun Shimizu<br>Shoji Tsuji<br>Tatsushi Toda | INTRODUCTION/AIMS: Heterozygous CGG repeat expansions in low-density lipoprotein receptor related protein 12 (LRP12) have recently been identified as a cause of oculopharyngodistal myopathy (OPDM), and the disease is designated as OPDM type 1 (OPDM1). In contrast to the broadening of our knowledge on the genetic background of OPDM, our knowledge of the clinical phenotype of genetically confirmed OPDM1 remains limited. | pmid:36052448<br>doi:10.1002/mus.27717 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 52 | pubmed:36052507 | Clinical, microbiological, and genomic characteristics of clade-III *Candida auris* colonization and infection in southern California, 2019-2022 | Annabelle de St Maurice<br>Urvashi Parti<br>Victoria E Anikst<br>Thomas Harper<br>Ruel Mirasol<br>Ayrton J Dayo<br>Omai B Garner<br>Kavitha K Prabaker<br>Shangxin Yang | CONCLUSIONS: We have demonstrated that a robust C. auris surveillance program can be established using both active and passive surveillance, with multidisciplinary efforts involving the microbiology laboratory and the hospital epidemiology team. In Los Angeles County, C. auris strains are highly related and echinocandins should be used for empiric therapy. | pmid:36052507<br>doi:10.1017/ice.2022.204 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 53 | pubmed:36052559 | Arsenic trioxide-loaded nanoparticles enhance the chemosensitivity of gemcitabine in pancreatic cancer *via* the reversal of pancreatic stellate cell desmoplasia by targeting the AP4/galectin-1 pathway | Yue Zhao<br>Hanming Yao<br>Kege Yang<br>Shiji Han<br>Shangxiang Chen<br>Yaqing Li<br>Shaojie Chen<br>Kaihong Huang<br>Guoda Lian<br>Jiajia Li | Pancreatic stellate cells (PSCs) constitute the fibrotic tumor microenvironment composed of the stroma matrix, which blocks the penetration of gemcitabine (GEM) in pancreatic adenocarcinoma (PDAC) and results in chemoresistance. We analyzed the expression of -SMA, collagen type I, and fibronectin by immunohistochemistry of pancreatic cancer tissues and demonstrated that the abundant interstitial stroma is associated with dismal survival. Two desmoplastic pancreatic tumor models are treated with... | pmid:36052559<br>doi:10.1039/d2bm01039a | Fri, 02 Sep 2022 06:00:00 -0400 |
| 54 | pubmed:36053007 | Carbon monoxide-releasing molecule-401, a water-soluble manganese-based metal carbonyl, suppresses *Prevotella intermedia* lipopolysaccharide-induced production of nitric oxide in murine macrophages | Eun-Young Choi<br>Jung Eun Lee<br>Ah Rim Lee<br>In Soon Choi<br>Sung-Jo Kim | CONCLUSION: The modulation of host inflammatory response by CORM-401 might be of help in the therapy of periodontal disease. | pmid:36053007<br>doi:10.1080/08923973.2022.2119998 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 55 | pubmed:36053129 | Antifungal therapies for allergic bronchopulmonary aspergillosis in people with cystic fibrosis | Natalie Z Francis<br>Kevin W Southern | BACKGROUND: Allergic bronchopulmonary aspergillosis (ABPA) is an allergic reaction to colonisation of the lungs with the fungus Aspergillus fumigatus, and affects around 10% of people with cystic fibrosis. ABPA is associated with an accelerated decline in lung function. High doses of corticosteroids are the main treatment for ABPA; although the long-term benefits are not clear, and their many side effects are well-documented. A group of compounds, the azoles, have activity against A fumigatus,... | pmid:36053129<br>doi:10.1002/14651858.CD002204.pub5 | Fri, 02 Sep 2022 06:00:00 -0400 |

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|---|---|---|---|---|---|---|
| 56 | pubmed:36053179 | Congenital lipoid adrenal hyperplasia in a Saudi infant | Siham Hussein Subki<br>Raghad Wadea Mohammed Hussain<br>Abdulmoein Eid Al-Agha | SUMMARY: Congenital lipoid adrenal hyperplasia (CLAH) is characterized by a defect in the STAR protein-encoding gene that attenuates all steroidogenesis pathways. Herein, we present the first reported case in Saudi Arabia of a 46 XY, phenotypically female infant with an unfamiliar, darkened complexion compared to the family's skin color. Based on the clinical and biochemical findings, CLAH was diagnosed and glucocorticoid replacement therapy was initiated. As a result, we suggest that... | pmid:36053179<br>doi:10.1530/EDM-22-0294 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 57 | pubmed:36053189 | Denovo DCHS1 splicing mutation in a patient with mitral valve prolapse | Dongping Li<br>Zhangqing Yi<br>Qin Wu<br>Yuyang Huang<br>Hanyi Yao<br>Zhiping Tan<br>Yifeng Yang<br>Weizhi Zhang | No abstract | pmid:36053189<br>doi:10.1093/qjmed/hcac214 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 58 | pubmed:36053203 | Clinical Trial Development in TP53-Mutated Locally Advanced and Recurrent/Metastatic Head and Neck Squamous Cell Carcinoma | Cristina P Rodriguez<br>Hyunseok Kang<br>Jessica L Geiger<br>Barbara Burtness<br>Christine H Chung<br>Curtis R Pickering<br>Carole Fakhry<br>Quynh Thu Le<br>Sue S Yom<br>Thomas J Galloway<br>Erica Golemis<br>Alice Li<br>Jeffrey Shoop<br>Stuart Wong<br>Ranee Mehra<br>Heath Skinner<br>Nabil F Saba<br>Elsa R Flores<br>Jeffrey N Myers<br>James M Ford<br>Rachel Karchin<br>Robert L Ferris<br>Charles Kunos<br>Jean M Lynn<br>Shakun Malik | TP53 mutation is the most frequent genetic event in head and neck squamous cell carcinoma (HNSCC), found in over 80% of patients with HPV-negative disease. As mutations in the TP53 gene are associated with worse outcomes in HNSCC, novel therapeutic approaches are needed for patients with TP53 mutated tumors. The National Cancer Institute (NCI) sponsored a Clinical Trials Planning Meeting (CTPM) to address the issues of identifying and developing clinical trials for patients with TP53 mutations.... | pmid:36053203<br>doi:10.1093/jnci/djac163 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 59 | pubmed:36053226 | Application of the Single-Molecule Real-Time Technology (SMRT) for Identification of HK Thalassemia Allele | Min Zhang<br>Zhaodong Lin<br>Meihuan Chen<br>Yali Pan<br>Yanhong Zhang<br>Lingji Chen<br>Na Lin<br>Yuanyuan Ren<br>Hongjin Jia<br>Meiying Cai<br>Liangpu Xu<br>Hailong Huang | CONCLUSION: The results demonstrate that SMRT offers a higher detection accuracy of thalassemia rare and new loci. It is an efficient, reliable, and broad-spectrum test that can be widely used for thalassemia screening in the clinic. | pmid:36053226<br>doi:10.1093/labmed/lmac065 | Fri, 02 Sep 2022 06:00:00 -0400 |

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|---|---|---|---|---|---|---|
| 60 | pubmed:36053258 | Sex-specific role of the circadian transcription factor NPAS2 in opioid tolerance, withdrawal and analgesia | Stephanie Puig<br>Micah A Shelton<br>Kelly Barko<br>Marianne L Seney<br>Ryan W Logan | Opioids like fentanyl remain the mainstay treatment for chronic pain. Unfortunately, opioid's high dependence liability has led to the current opioid crisis, in part, because of side-effects that develop during long-term use, including analgesic tolerance and physical dependence. Both tolerance and dependence to opioids may lead to escalation of required doses to achieve previous therapeutic efficacy. Additionally, altered sleep and circadian rhythms are common in people on opioid therapy.... | pmid:36053258<br>doi:10.1111/gbb.12829 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 61 | pubmed:36053276 | Small-molecule inhibition of the acyl-lysine reader ENL as a strategy against acute myeloid leukemia | Yiman Liu<br>Qinglan Li<br>Fatemeh Alikarami<br>Declan R Barrett<br>Leila Mahdavi<br>Hangpeng Li<br>Sylvia Tang<br>Tanweer A Khan<br>Mayako Michino<br>Connor Hill<br>Lele Song<br>Lu Yang<br>Yuanyuan Li<br>Sheela Pangeni Pokharel<br>Andrew W Stamford<br>Nigel Liverton<br>Louis M Renzetti<br>Simon Taylor<br>Gillian F Watt<br>Tammy Ladduwahetty<br>Stacia Kargman<br>Peter T Meinke<br>Michael A Foley<br>Junwei Shi<br>Haitao Li<br>Martin Carroll<br>Chun-Wei Chen<br>Alessandro Gardini<br>Ivan Maillard<br>David J Huggins<br>Kathrin M Bernt<br>Liling Wan | The chromatin reader eleven-nineteen-leukemia (ENL) has been identified as a critical dependency in AML, but its therapeutic potential remains unclear. We describe a potent and orally bioavailable small-molecule inhibitor of ENL, TDI-11055, which displaces ENL from chromatin by blocking its YEATS domain interaction with acylated histones. Cell lines and primary patient samples carrying MLL rearrangements or NPM1 mutations are responsive to TDI-11055. A CRISPR-Cas9-mediated mutagenesis screen... | pmid:36053276<br>doi:10.1158/2159-8290.CD-21-1307 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 62 | pubmed:36053497 | Musicians Focal Dystonia: The Practitioner's Perspective on Psychological, Psychosocial, and Behavioural Risk Factors and Non-motor Symptoms | Anna Détári<br>Hauke Egermann | Musician's focal dystonia (MFD) is a painless, task-specific neurological movement disorder that impairs fine motor control when playing an instrument. The pathophysiology is not fully understood, and while the available treatment strategies can help with improving motor control, they are rarely able to fully and reliably rehabilitate playing skills. Recent studies suggest that apart from genetic factors, maladaptive neuroplasticity, and the repetitive nature of the instrumental technique,... | pmid:36053497<br>doi:10.21091/mppa.2022.3023 | Fri, 02 Sep 2022 06:00:00 -0400 |

| | NCT Number | Title | Authors | Description | Identifier | Dates |
|---|---|---|---|---|---|---|
| 63 | pubmed:36053672 | A bone morphogenetic protein signaling inhibitor, LDN193189, converts ischemia-induced multipotent stem cells into neural stem/progenitor cell-like cells | Yusuke Minato<br>Akiko Nakano-Doi<br>Seishi Maeda<br>Takayuki Nakagomi<br>Hideshi Yagi | Stem cell therapy is used to restore neurological function in stroke patients. We have previously reported that ischemia-induced multipotent stem cells (iSCs), which are likely derived from brain pericytes, develop in post-stroke human and mouse brains. Although we have demonstrated that iSCs can differentiate into neural lineage cells, the factors responsible for inducing this differentiation remain unclear. In this study, we found that LDN193189, a BMP inhibitor, caused irreversible changes in... | pmid:36053672<br>doi:10.1089/scd.2022.0139 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 64 | pubmed:36053930 | LINC00536 knockdown inhibits breast cancer cells proliferation, invasion, and migration through regulation of the miR-4282/centromere protein F axis | Mei-Feng Zhou<br>Wei Wang<br>Lin Wang<br>Jin-Dian Tan | Breast cancer (BC) poses a huge threat to women's health. Growing evidence has shown lncRNAs play critical roles in BC progression. However, the effect of LINC00536 in BC remains unknown. LINC00536, miR-4282, and centromere protein F (CENPF) expressions in BC cells were determined using qPCR assay. Colony formation assay was employed to evaluate the cell proliferation of BC cells. Besides, cell migration and invasion were evaluated using the transwell assay. FISH assay was employed to analyze... | pmid:36053930<br>doi:10.1002/kjm2.12583 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 65 | pubmed:36053975 | Effector Th1 cells under PD-1 and CTLA-4 checkpoint blockade abrogate the upregulation of multiple inhibitory receptors and by-pass exhaustion | Utku Horzum<br>Hamdullah Yanik<br>Ekim Z Taskiran<br>Gunes Esendagli | Immune checkpoint inhibitor (ICI) immunotherapy relies on the restoration of T-cell functions. The ICI receptors are not only found on exhausted T cells but also upregulated upon activation and reach high levels on effector T cells. In an ex vivo model, this study explored the consequences of PD-1 and cytotoxic T-lymphocyte antigen (CTLA-4) blockade applied during specific time frames of T-cell stimulation that coincide with distinct functional phases in type 1 helper T (Th1) cells. When applied... | pmid:36053975<br>doi:10.1111/imm.13560 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 66 | pubmed:36054236 | C allele in transforming growth factor-1 rs1800471 gene polymorphisms might indicate a protective feature in encapsulating peritoneal sclerosis development | Feyza Bora<br>Bengisu Aslan<br>Funda Sar<br>Fatih Ylmaz<br>Fettah Fevzi Ersoy<br>Sadi Köksoy<br>Sebahat Özdem<br>kbal Özen Küçükçetin<br>Murat Sipahiolu<br>brahim Karakaya<br>Yener Koç<br>Memnune Sena Ulu | CONCLUSION: C allele in TGF-1 rs1800471 gene polymorphisms might indicate a protective feature in EPS development. Knowing the presence of polymorphism may be effective in selecting renal replacement therapy in patients. | pmid:36054236<br>doi:10.1111/1744-9987.13913 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 67 | pubmed:36054621 | Evaluation and Management of Hypoparathyroidism Summary Statement and Guidelines from the Second International Workshop | Aliya A Khan<br>John P Bilezikian<br>Maria Luisa Brandi<br>Bart L Clarke<br>Neil J Gittoes<br>Janice L Pasieka<br>Lars Rejnmark<br>Dolores M Shoback<br>John T Potts<br>Gordon H Guyatt<br>Michael Mannstadt | This clinical practice guideline addresses the prevention, diagnosis and management of hypoparathyroidism (HypoPT) and provides evidence-based recommendations. The HypoPT Task Forces included 4 teams with a total of 50 international experts including representatives from the sponsoring societies. A methodologist (GG) and his team supported the taskforces and conducted the systematic reviews. A formal process following GRADE methodology and the systematic reviews provided the structure for 7 of... | pmid:36054621<br>doi:10.1002/jbmr.4691 | Fri, 02 Sep 2022 06:00:00 -0400 |

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| 68 | pubmed:36054650 | Adalimumab therapy is associated with increased faecal short chain fatty acids in hidradenitis suppurativa | Artiene Tatian<br>Sara Bordbar<br>Samuel Der Sarkissian<br>Jane A Woods<br>Geoffrey D Cains<br>Chun Wie Chong<br>Eliana Mariño<br>John W Frew | Altered gut microbiota composition has been observed in individuals with hidradenitis suppurutiva (HS) and many other inflammatory diseases, including obesity, type 1 and type 2 diabetes. Here, we addressed whether adalimumab, a systemic anti-inflammatory therapy, may impact the microbiota biochemical profile, particularly on beneficial metabolites such as short-chain fatty acids (SCFAs). We conducted an observational single-arm pilot trial to assess gut microbiota composition by 16S rRNA gene... | pmid:36054650<br>doi:10.1111/exd.14665 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 69 | pubmed:36054652 | Pulmonary Delivery of Levamisole Nanoparticles as an Immunomodulator Affecting Th and a Potential ADAM10 Inhibitor to Ameliorate Severe Allergic Asthma | Liping Fang<br>Nasser Nikfarjam<br>Mohammad Gharagozlou<br>Tao Huang<br>Yu Song<br>Ziba Islambulchilar<br>Abdolreza Esmaeilzadeh<br>Davood Jafari<br>Seyyed Shamsadin Athari | Asthma is a common chronic lung disease without absolute treatment, and hypersensitivity reactions and type 2 immune responses are responsible for asthma pathophysiology. ADAM10 as a metalloproteinase transmembrane protein is critical for development of Th2 responses, and levamisole as an anthelmintic drug has immunomodulatory effects, which not only regulates ADAM10 activity but also can suppress the bone marrow and neutrophil production. Therefore, in the present study, nanoparticles were used... | pmid:36054652<br>doi:10.1021/acsbiomaterials.2c00843 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 70 | pubmed:36054749 | Level of therapeutic innovation from the registration studies of the new drugs for the prophylaxis of migraine | Domenico Motola<br>Greta Santi Laurini<br>Giulia Bonaldo<br>Nicola Montanaro | WHAT IS KNOWN AND OBJECTIVE: Migraine is one of the most prevalent and disabling medical illnesses. Preventive drugs are used to reduce the frequency, severity, and duration of attacks. Most patients were no longer on their medication due to contraindications or poor clinical response. Therefore, there is need for novel prophylactic agents for migraine. New preventive treatments are those of the class of calcitonin gene related peptide (CGRP)-targeting therapies. We aimed to assess the real... | pmid:36054749<br>doi:10.1111/jcpt.13760 | Fri, 02 Sep 2022 06:00:00 -0400 |
| 71 | pubmed:36054863 | Update on spinal muscular atrophy treatment | Ricardo Erazo Torricelli | Spinal muscular atrophy (SMA) has been known as a clinical entity for 130 yearsis still recognized today as the most severe autosomal recessive neuromuscular disease (5q,13,2) in pediatrics. Until 2015, SMA treatment was limited to ventilatory, nutritional, and physical therapy support. Currently, the existence of genetic treatments: gene modification by inclusion of exon 7 to the SMN2 gene (nusinersen and risdiplam) or insertion of the SMN1 gene through the adeno-associated viral transporter... | pmid:36054863 | Fri, 02 Sep 2022 06:00:00 -0400 |

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| 72 | pubmed:36054881 | Clinical and Molecular Determinants of Clonal Evolution in Aplastic Anemia and Paroxysmal Nocturnal Hemoglobinuria | Carmelo Gurnari<br>Simona Pagliuca<br>Pedro Henrique Prata<br>Jacques-Emmanuel Galimard<br>Luiz Fernando B Catto<br>Lise Larcher<br>Marie Sebert<br>Vincent Allain<br>Bhumika J Patel<br>Arda Durmaz<br>Andre L Pinto<br>Mariana C B Inacio<br>Lucie Hernandez<br>Nathalie Dhedin<br>Sophie Caillat-Zucman<br>Emmanuelle Clappier<br>Flore Sicre de Fontbrune<br>Maria Teresa Voso<br>Valeria Visconte<br>Régis Peffault de Latour<br>Jean Soulier<br>Rodrigo T Calado<br>Gérard Socié<br>Jaroslaw P Maciejewski | CONCLUSION: The risk of sMN in AA is associated with disease severity, lack of response to treatment, and patients' age. sMNs display high-risk morphological, karyotypic, and molecular features. The landscape of acquired somatic mutations is complex and incompletely understood and should be considered with caution in medical management. | pmid:36054881<br>doi:10.1200/JCO.22.00710 | Fri, 02 Sep 2022 06:00:00 -0400 |