Resource Summary Report

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Phelan-McDermid Syndrome Foundation

RRID:SCR_001707
Type: Tool

Proper Citation

Phelan-McDermid Syndrome Foundation (RRID:SCR_001707)

Resource Information

URL: https://pmsf.org/

Proper Citation: Phelan-McDermid Syndrome Foundation (RRID:SCR_001707)

Description: The Phelan-McDermid Syndrome Foundation, established in 2002, is a 501(c)3 nonprofit group that provides support services for those who have family members affected by 22q13 Deletion Syndrome / Phelan-McDermid Syndrome. It also raises money to further awareness of the syndrome through research and sponsoring an international conference every two years that brings together families, researchers and therapists. The Foundation facilitates connections between families through networking, communications and support services. We also build alliances with other rare diseases groups to expand our reach and exposure. The syndrome, which affects families worldwide, is a rare genetic occurrence and is the result of a damaged or missing protein on the 22nd chromosome. Our Foundation works with researchers who are looking into the cause and possible cure for the syndrome. PMSF's grants and fellowships program is intended to encourage research projects that will advance the development of treatments and cures for PMS. Our mission is to bring together everyone affected by 22q13 Deletion Syndrome/Phelan-McDermid Syndrome to help them through the challenges they face every day and to raise awareness in the medical and research communities.

Abbreviations: PMSF

Resource Type: community building portal, data or information resource, disease-related portal, funding resource, portal, topical portal

Keywords: 22q13 deletion syndrome, phelan-mcdermid syndrome, rare disease, genetic, meeting, child, chromosome 22, treatment, therapy, research, grant, fellowship

Related Condition: Phelan-McDermid Syndrome

Availability: Free, Freely Available

Resource Name: Phelan-McDermid Syndrome Foundation

Resource ID: SCR_001707

Alternate IDs: nif-0000-10203

Record Creation Time: 20220129T080209+0000

Record Last Update: 20260905T132438+0000

Ratings and Alerts

No rating or validation information has been found for Phelan-McDermid Syndrome Foundation.

No alerts have been found for Phelan-McDermid Syndrome Foundation.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 33 mentions in open access literature.

Listed below are recent publications. The full list is available at NIF .

Wu X, et al. (2025) Identification of a cryptic unbalanced translocation Der(22)t(12;22)(q24.33;q13.33) in a large Chinese family with Phelan-McDermid syndrome by nanopore sequencing. Scientific reports, 15(1), 2656.

Hao Y, et al. (2022) Prenatal and postnatal diagnosis of Phelan-McDermid syndrome: A report of 21 cases from a medical center and review of the literature. Frontiers in genetics, 13, 961196.

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Matesanz N, et al. (2017) MKK6 controls T3-mediated browning of white adipose tissue. Nature communications, 8(1), 856.

Zhou X, et al. (2017) Overexpression of MPC1 inhibits the proliferation, migration, invasion, and stem cell-like properties of gastric cancer cells. OncoTargets and therapy, 10, 5151.

Andres-Mach M, et al. (2017) A Long-Term Treatment with Arachidonyl-2'-Chloroethylamide Combined with Valproate Increases Neurogenesis in a Mouse Pilocarpine Model of Epilepsy. International journal of molecular sciences, 18(5).

Gaullier G, et al. (2016) A higher-order entity formed by the flexible assembly of RAP1 with TRF2. Nucleic acids research, 44(4), 1962.

Xiao X, et al. (2015) Synthetic A?? peptides acquire prion-like properties in the brain. Oncotarget, 6(2), 642.

Pellegatta S, et al. (2015) Effective immuno-targeting of the IDH1 mutation R132H in a murine model of intracranial glioma. Acta neuropathologica communications, 3, 4.

Papp SJ, et al. (2015) DNA damage shifts circadian clock time via Hausp-dependent Cry1 stabilization. eLife, 4.

Xue M, et al. (2015) Distinct patterns of the histone marks associated with recruitment of the methionine chain-elongation pathway from leucine biosynthesis. Journal of experimental botany, 66(3), 805.

Sharma B, et al. (2015) Mycobacterium tuberculosis secretory proteins downregulate T cell activation by interfering with proximal and downstream T cell signalling events. BMC immunology, 16, 67.

Mercenne G, et al. (2015) Angiomotin functions in HIV-1 assembly and budding. eLife, 4.

Pekkonen P, et al. (2014) KSHV viral cyclin interferes with T-cell development and induces lymphoma through Cdk6 and Notch activation in vivo. Cell cycle (Georgetown, Tex.), 13(23), 3670.

Jiang YS, et al. (2014) Probucol suppresses human glioma cell proliferation in vitro via ROS production and LKB1-AMPK activation. Acta pharmacologica Sinica, 35(12), 1556.

Su YF, et al. (2014) A putative novel protein, DEPDC1B, is overexpressed in oral cancer patients, and enhanced anchorage-independent growth in oral cancer cells that is mediated by Rac1 and ERK. Journal of biomedical science, 21(1), 67.

Booth DS, et al. (2014) The export receptor Crm1 forms a dimer to promote nuclear export of HIV RNA. eLife, 3, e04121.

Siniscalco D, et al. (2014) The in vitro GcMAF effects on endocannabinoid system transcriptionomics, receptor formation, and cell activity of autism-derived macrophages. Journal of neuroinflammation, 11, 78.

Zhang M, et al. (2013) Overexpression of JAM-A in non-small cell lung cancer correlates with tumor progression. PloS one, 8(11), e79173.

Kangussu-Marcolino MM, et al. (2013) Distinct genomic organization, mRNA expression and cellular localization of members of two amastin sub-families present in Trypanosoma cruzi. BMC microbiology, 13, 10.