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RESEARCH AREA

1/ Our goal is to understand the role of DNA methylation in both physiological and pathological conditions. We work with conditional knockout mice to elucidate the roles of de novo methyltransferases (DNMT3a and b) in embryonic development, differentiation, maturation, and aging. We employ epigenetic methods to characterize our models.

2/ Ectopic calcification is characteristic of various rare and common diseases, such as chronic kidney disease. Loss-of-function mutations in different proteins (e.g., ABCC6) lead to rare hereditary diseases characterized by ectopic mineralization. Recently, we have identified variants causing incomplete penetrance diseases. Currently, we aim to understand the pathophysiological roles of the disease-causing proteins and epigenetic mechanisms regulating ectopic calcification processes.

TECHNIQUES AVAILABLE IN THE LAB

Our research group's main focus is conducting epigenetic studies. We perform DNA methylation analysis using next-generation sequencing, study mammalian cell cultures and mouse models, analyze proteins and epigenetic modifications through Western blotting, and carry out gene expression analysis using qPCR.

SELECTED PUBLICATIONS

Jain, P., Miller-Fleming, T., Topaloudi, A, Yu, D., Drineas, P., Georgitsi, M., Yang, Z., Rizzo, R., Müller-Vahl, KR., Tumer, Z., Mol Debes, N., Hartmann, A., Depienne, C., Worbe, Y., Mir, P., Cath, DC., Boomsma, DI., Roessner, V., Wolanczyk, T., Janik, P., Szejko, N., Zekanowski, C., Barta, C., Nemoda, Z., Tarnok, Z., Buxbaum, JD., Grice, D., Glennon, J., Stefansson, H., Hengerer, B., Benaroya-Milshtein, N., Cardona, F., Hedderly, T., Heyman, I., Huyser, C., Morer, A., Mueller, N., Munchau, A., Plessen, KJ., Porcelli, C., Walitza, S., Schrag, A., Martino, D.; Psychiatric Genomics Consortium Tourette Syndrome Working Group (PGC-TS); EMTICS collaborative group; Dietrich A; **TS-EUROTRAIN Network**; Mathews CA, Scharf JM, Hoekstra PJ, Davis LK, Paschou P. (2023). Polygenic risk score-based phenome-wide association study identifies novel association for Tourette syndrome. *Transl Psychiatry* **13**(1): 69.

Tsetsos, F., Topaloudi, A., Jain, P., Yang, Z., Yu, D., Kolovos, P., Tumer, Z., Rizzo, R., Hartmann, A., Depienne, C., Worbe, Y., Müller-Vahl, KR., Cath, DC., Boomsma, DI., Wolanczyk, T., Zekanowski, C., Barta, C., Nemoda, Z., Tarnok, Z., Padmanabhuni, SS., Buxbaum, JD., Grice, D., Glennon, J., Stefansson, H., Hengerer, B., Yannaki, E., Stamatoyannopoulos, JA., Benaroya-Milshtein, N., Cardona, F., Hedderly, T., Heyman, I., Huyser, C., Mir, P., Morer, A., Mueller, N., Munchau, A., Plessen, KJ., Porcelli, C., Roessner, V., Walitza, S., Schrag, A., Martino, D.; PGC TS Working Group; TSAICG; TSGeneSEE Initiative; EMTICS Collaborative Group; **TS-EUROTRAIN Network**; TIC Genetics Collaborative Group; Tischfield JA, Heiman GA, Willsey AJ, Dietrich A, Davis LK, Crowley JJ, Mathews CA, Scharf JM, Georgitsi M, Hoekstra PJ, Paschou P. (2023) Genome-wide association study points to novel locus for Gilles de la Tourette syndrome. *Biol Psychiatry Online* ahead of print.

Liang, X., **Aranyi, T.**, Zhou, J., Guan, Y., Liu, H., Susztak, K., (2023) Tet2 and Tet3 mediated cytosine hydroxymethylation in Six2 progenitor cells is critical for nephron progenitor differentiation and nephron endowment. *J Am Soc Nephrol* **34**(4): 572-89.

Szeri, F., Miko, A., Navasiolava, N., Kaposi, A., Verschuere, S., Molnar, B., Li, Q., Terry, SF., Boraldi, F., Uitto, J., van de Wetering, K., Martin, L., Quaglino, D., Vanakker, OM., Tory, K., **Aranyi, T.*** (2022) The pathogenic c.1171A>G (p.Arg391Gly) and c.2359G>A (p.Val787Ile) ABCC6 variants display incomplete penetrance causing pseudoxanthoma elasticum in a subset of individuals. *Hum Mutat* **43**(12): 1872-81.

Belal, S., Goudenège, D., Bocca, C., Dumont, F., Chao De La Barca, JM., Desquiret-Dumas, V., Gueguen, N., Geffroy, G., Benyahia, R., Kane, S., Khiati, S., Bris, C., **Aranyi, T.**, Stockholm, D., Inisan, A., Renaud, A., Barth, M., Simard, G., Reynier, P., Letournel, F., Lenaers, G., Bonneau, D., Chevrollier, A., Procaccio, V. (2022) Glutamate-Induced Dereglulation of Krebs Cycle in Mitochondrial Encephalopathy Lactic Acidosis Syndrome Stroke-Like Episodes (MELAS) Syndrome Is Alleviated by Ketone Body Exposure. *Biomedicines* **10** (7): 1665.