**CZI Channelopathy Library**

**Thursday, 12/12/2024**

**Sources used for this (also linked in my Zotero Library):**

1. Kim, J.-B. Channelopathies. *Korean J. Pediatr.* 57, 1–18 (2014).

2. Sahin-Tóth, M. Channelopathy of the Pancreas Causes Chronic Pancreatitis. *Gastroenterology* 158, 1538–1540 (2020).

3. Wang, J. *et al.* Epilepsy-associated genes. *Seizure* 44, 11–20 (2017).

4. Hildebrandt, F. Genetic kidney diseases. *Lancet* 375, 1287–1295 (2010).

5. Guerrini, R., Balestrini, S., Wirrell, E. C. & Walker, M. C. Monogenic Epilepsies. *Neurology* 97, 817–831 (2021).

6. Imbrici, P. *et al.* Therapeutic Approaches to Genetic Ion Channelopathies and Perspectives in Drug Discovery. *Front. Pharmacol.* 7, (2016).

7. fphar-07-00121-g001.jpg (1950×1352). https://www.frontiersin.org/files/Articles/195106/fphar-07-00121-HTML/image\_m/fphar-07-00121-g001.jpg.

**Link to Google Sheets gene list. Currently located in Schaffer Lab Google Drive:**

https://docs.google.com/spreadsheets/d/12-6vNZiYNigvZ0Pe-9HsWZAbab4EtLPiVby4ilhTOrU/edit?usp=sharing

**Uniprot Scraper Python Script (Written by Connor Taseff):** https://github.com/ctaseff02/uniprot-scraper

**Databases/Resources Used:**

UniProt

Ensembl Variant Effect Predictor

**PolyPhen Scoring Index:** https://ionreporter.thermofisher.com/ionreporter/help/GUID-57A60D00-0654-4F80-A8F9-F6B6A48D0278.html

**Workflow:**

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Pulled Genes from literature

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Found on Uniprot and ran through attached scraper script to pull out Pathogenic, Likely Pathogenic, or VUS variants

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Script called to the Ensembl VEP API and pulled out the PolyPhen scores for all missense variants

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Selected all variants with "Likely pathogenic" and "Probably pathogenic" scores

●

All final variant lists are moved into the Google Sheet document. Raw data available as Excel spreadsheets in ZIP file on my computer/in my email