TREATABOLOME DATABASES FOR ACCELERATED DISCOVERY, TESTING AND IMPLEMENTATION OF VARIANT-SPECIFIC THERAPIES FOR RARE GENETIC NEURODEVELOPMENTAL DISEASES

Several recent publications draw attention to two databases for the treatment of rare, Mendelian metabolic diseases that cause developmental delay. These initiatives show that a precise genetic diagnosis can result in an equally precise clinical intervention by nutritional, pharmacological or vitamin/trace element supplementation therapies. These are simple and surprisingly effective in the majority of cases, much more so than the much more expensive enzyme replacement therapies.
The first is from the Treatabolome and International Rare Diseases Research Consortium (www.treatable-id.org), see the article by Hoytema van Konijnenburg in the Orphanet Journal of Rare Diseases. The database exists since 2012 and has now been updated. It provides therapeutic information on 116 disorders (with 139 genes involved).
A second is from the International Rare Diseases Research Consortium (Solve-RD, www.solve-rd.eu), founded in 2011. The database and its history are described in several papers in the Journal of Neuromuscular diseases issue of May 13, 2021, see the introductory paper by Bonne. The database covers gene variants causing Parkinson's disease, skeletal muscle ion channelopathies, peripheral neuropathies, metabolic myopathies related to glycogen storage and lipid metabolism, and laminopathies, all with the aim to have 1000 new treatment options of proven effectiveness by 2027.

Using these databases, clinicians have easy access to trustable, up-to-date, evidence-based information for the timely treatment of patients with such rare diseases.

Links:
https://ojrd.biomedcentral.com/articles/10.1186/s13023-021-01727-2
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC8203244/pdf/jnd-8-jnd219003.pdf

Lorenzo's oil:
https://en.wikipedia.org/wiki/Lorenzo%27s_Oil