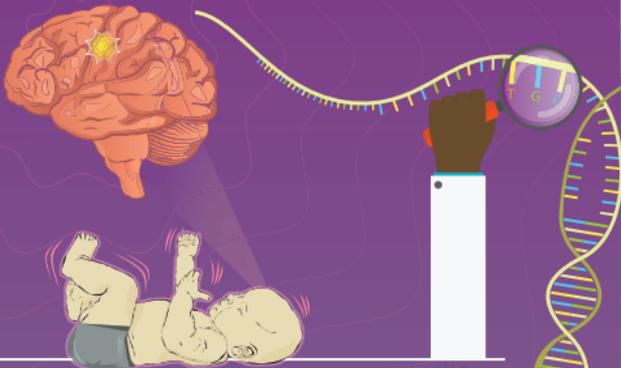


Whole-Genome Sequencing for Diagnosing Developmental Epileptic Encephalopathies

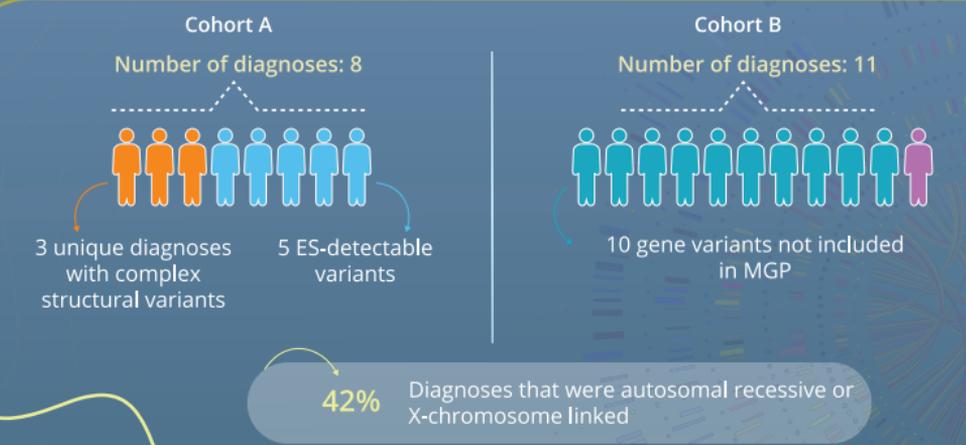
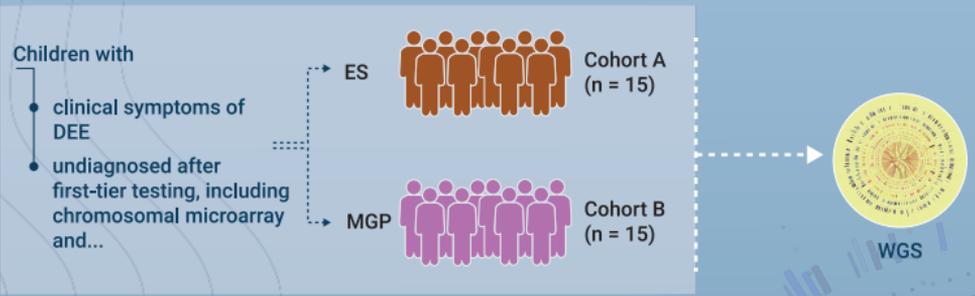
Developmental and epileptic encephalopathies (DEE) are a group of disorders characterized by uncontrolled seizures that have an impact on childhood development



Exome sequencing (ES) and multigene panel (MGP) testing have been extensively used for molecular diagnosis of DEE

Study question

Can whole-genome sequencing (WGS) improve the diagnostic yield in patients with DEE?



WGS improves diagnostic yield in patients with DEE by detecting complex structural variants