Fly’s genome helps identify genes involved in human neurological diseases

The combination of fly genetics and genomics with human genome knowledge enables rapid identification of human disease-causing mutations, including those that result in rare single gene or Mendelian disorders, said an international consortium of researchers led by those at Baylor College of Medicine and the Jan and Dan Duncan Neurological Research Institute at Texas Children’s Hospital.

“Usually, mapping chemically induced mutations is like searching for a needle in the haystack and is the major roadblock in mapping mutations obtained from large-scale fly screens. Combining coarse mapping with whole genome sequencing greatly facilitates this endeavor,” said Hugo Bellen, professor of molecular and human genetics, member of the faculty of the Neurological Research Institute and director of the Baylor College of Medicine Program in Developmental Biology. He also is a Howard Hughes Medical Institute investigator.

The scientists then determined how this information about the fly genes applied to the human genome by identifying and comparing their human counterpart genes (homologs) for variations. They used human exome (the protein-coding part of the genome) databases created by genomic sequencing of DNA samples from families with neurologic disease traits. These series of steps allowed them to quickly pinpoint specific disease-causing mutations in many known and novel human genes.

“It was astonishing to see how quickly basic biological and genomic information obtained from flies could be employed to identify molecular lesions responsible for rare genetic disorders,” said Dr. Manish Jaiswal, a postdoctoral fellow in the Bellen lab and co-first author of the report.

Read full original article: Researchers harness power of fly genetics, genomics to speed identification of human neurological disease genes