HDL2 iPSCs cell line

Catalogue number: 156417
Sub-type:
Images:

Contributor

Inventor: Russell Margolis
Institute: Johns Hopkins University
Images:

Tool details

*FOR RESEARCH USE ONLY

Name: HDL2 iPSCs cell line

Alternate name:

Class:
Conjugate:
Description: This cell line was generated from induced pluripotent stem cells derived from fibroblast cell lines developed from patients with Huntington's disease-like 2. HDL2, discovered and genetically defined by the Margolis group, is a rare, autosomal dominant neurodegenerative disorder, clinically and neuropathologically very similar to HuntingtonÄ?Ë???Â???Â?s disease. Like HD, the neuropathology of HDL2 is characterized by cortical and striatal neurodegeneration and the presence of neuronal protein aggregates. HDL2 is caused by a CTG/CAG expansion on chromosome 16q24. Normal alleles contain 6-28 triplets, while pathogenic repeats range from 40-59 triplets, remarkably similar to HD. In the CTG orientation, the repeat falls in the gene junctophilin-3 (JPH3). The development of HDL2 iPS cells as a tool for studying HDL2 provides several important opportunities including; an improved understanding of HDL2 itself,new insights into fundamental pathogenic processes relevant to other repeat expansion diseases, andthe opportunity to find pathogenic points of convergence between HD and HDL2 that will lead to a focus on therapeutic targets of most promise for both diseases.
Purpose:
Parental cell:
Organism:
Tissue:
Model: Stem Cells
Gender:
Isotype:
Reactivity:
Selectivity:

Host:
Immunogen:
Immunogen UNIPROT ID:
Sequence:
Growth properties:
Production details:
Formulation:
Recommended controls:
Bacterial resistance:
Selectable markers:
Additional notes:

Target details

Target: Huntington's disease-like 2 (HDL2)

Target alternate names:

Target background:

Molecular weight:

Ic50:

Applications

Application:
Application notes:

Handling

Format: Frozen
Concentration:
Passage number:
Growth medium:
Temperature:
Atmosphere:
Volume:
Storage medium:
Storage buffer:
Storage conditions:
Shipping conditions: Dry ice

Related tools

Related tools:

References

References: