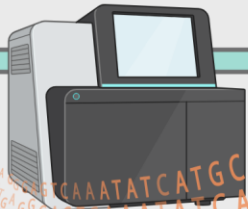


Initial Assessment



Clinical examination



Routine genetic/genomic test followed by bioinformatics and genetic analysis



Genetic report

Precision Diagnostic Unit

Clinicians from HSJD clinical services, collaborators, and external entities propose an undiagnosed patient to the PGF-SJD specialists after a deep phenotyping

Decision of patient inclusion based on the PGF-SJD inclusion criteria by clinicians and the PGF-SJD core team

Functional test request through the hospital PGF-SJD platform **DTSILICO, DTDNA, DTRNA & DTFIBR**



DTSILICO STUDY

Phase 1

Analysis of the phenotype and the provided genetic information

Phase 2

Analysis of the variant's pathogenicity

Phase 3

In-depth genomic analysis with deep-learning

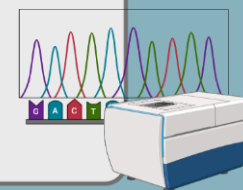
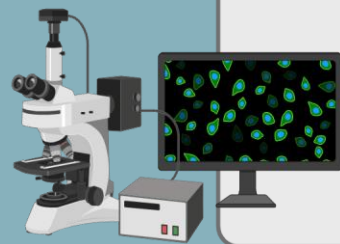
Yes

No

Sample collection Recommendation

Experimental procedures: DTDNA / DTRNA / DTFIBR

Gene expression, splicing events and protein expression, subcellular localization, function and interaction



Functional or biological report **uploaded to the hospital platform**

Diagnosis

Discussion and diagnostic decision-making by clinicians

Final genetic diagnosis

No genetic diagnosis and case re-evaluation