

GEO
Datasets

HD2
n=3,3

FTD
n=4,4

HD
n=7,7

DS
n=11,9

DM
n=8,21

ASD
n=12,12

Preprocessing

Quality
Assessment
(FASTQC)

Quality filtering
(trimmomatic)

Alignment
(STAR)

Differential
Splicing
Analysis with the
6 AS Tools

rMATS

Count Reads

MAJIQ build

BAM to Junc

Salmon
Index

Generate
Events

Whippet
Index

DS and Exon
Usage Analysis

MAJIQ
Quant

Intron
Clustering

Salmon
Quant

Whippet
Quant

Voila

Differential
Exon Excision
Analysis

PSI Per
Event

Whippet
Delta

rMATS
output

DEXSeq
output

MAJIQ
output

leafcutter
output

SUPPA2
output

whippet
output

Conversion of AS tool outputs to
standardised outputs

AS Tool
Comparison

AS Tool Output Comparison:
DS Gene overlap, DS Event overlap, dPSI comparison
between overlapping events