

# CORTAR

Relative quantification of pre-mRNA splicing from RNA-seq

## 281\_\_MTHFR\_\_P

**Significantly different splicing events (>2SD) compared to 2 controls.**

**Details:**

- RNA-seq: AGRF rRNA-depletion
- Controls: 2 disease-controls run in same flow cell
- Report: 2022-04-20
- Version: v0.3.0

Events marked with an asterisk (\*) are unique to the proband and any family members included in the analysis, as compared to controls. These events may be present at very low levels in the population.

### Overview

In the proband, MTHFR has pre-mRNA splicing events which are significantly different to controls (>5% change, >2 standard deviations) in 3 introns. All pre-mRNA splicing events occurring in these affected introns are highlighted below.

#### intron 6

chr:start-end	event	proband	controls	difference	frame
chr1:11794864-11795097	reduced normal exon 6-7	53.03%	100.00%	-46.97%	TRUE
chr1:11794539-11795097	increased exon 7 skipping*	42.42%	0.00%	42.42%	TRUE
chr1:11794864-11796205	increased exon 6 skipping*	4.55%	0.00%	4.55%	FALSE

#### intron 7

chr:start-end	event	proband	controls	difference	frame
chr1:11794539-11794728	reduced normal exon 7-8	17.92%	58.56%	-40.64%	TRUE
chr1:11794539-11795097	increased exon 7 skipping*	20.07%	0.00%	20.07%	TRUE
chr1:11794463-11794728	increased cryptic acceptor use*	18.64%	0.00%	18.64%	FALSE
chr1:11794539-11794728	increased intron 7 retention	43.37%	37.44%	5.92%	

## intron 9

chr:start-end	event	proband	controls	difference	frame
chr1:11792380-11793906	reduced normal exon 9-10	77.82%	86.09%	-8.26%	TRUE
chr1:11792380-11793906	increased intron 9 retention	20.62%	13.72%	6.91%	
chr1:11792380-11794357	increased exon 9 skipping*	1.56%	0.00%	1.56%	FALSE