Identity

Other names: RecQ4; RTS; RecQ protein like 4; ATP-dependent DNA helicase Q4
HGNC (Hugo): RECQL4
Location: 8q24.3

DNA/RNA

Description
Spans 6,46 kb; 21 exons; helicase domain is encoded by exons from 8 to 14.

Transcription
3,62 kb mRNA.

MUT 1
1650del17
Early stop codon 14 bp downstream

MUT 2
C2269T
nonsense

MUT 3
2492del2
Early stop codon

MUT 4
3’ splice site
Leds to skipping of exon 13

MUT 5
1573delT
Early stop codon 97 bp downstream

MUT 6
3’ splice site
G>A
mRNA del 93 bp

RecQ4 product and mutations

Helicase domain
Protein

**Description**
1208 aa; 13,3 kDa; belongs to the RecQ subfamily of helicases and contains from aa 476 to 824 an helicase domain with a potential ATP binding site from aa 502 to 509, and the DEAH box from aa 605 to 608.

**Expression**
The RecQ4 gene is predominantly expressed in thymus and testis and at low levels in other organs such as heart, brain, placenta, pancreas, small intestine, and colon, indicating that the expression of RecQ4 gene is somewhat tissue-specific. The overall expression profile resembles that of the BLM gene. Interestingly, the expression of RecQ4 gene is partially upregulated in the G1/S phase of cell cycle.

**Localisation**
Nuclear.

**Function**
Suppresses promiscuous genetic recombination and ensures accurate chromosome segregation.

**Homology**
WRN/RECQ3, BLM/RECQ2.

**Mutations**

**Germinal**
See diagram of loss-of-function mutations in Rothmund-Thomson Syndrome patients.

Implicated in

**Rothmund-Thomson Syndrome**

**Disease**
Autosomal recessive disorder associated with genomic instability, cancer predisposition and premature ageing.

**References**


This article should be referenced as such: