COMING SOON

gb PHARM NUDT15 (R139C)

Clinical implications

NUDT15 gene encodes an enzyme that belongs to the nudix hydrolase superfamily, which catalyse the hydrolysis of deoxynucleoside diphosphates and triphosphates. The NUDT15 enzyme is directly involved in the metabolism of thiopurines. Individuals with reduced or absent NUDT15 activity have increased sensitivity to thiopurines, including and increased risk of severe myelosuppresion.

The no-function NUDT15*3 allele is characterized by a missense variant in exon 3 (NM_018283.4:c.415C>T, p.Arg139Cys, rs116855232). This amino acid change results in a nearly complete loss of enzymatic activity and protein stability in vitro. The no-function NUDT15*2 allele is characterized by a missense variant in exon 3 (NM_018283.4:c.415C>T, p.Arg139Cys, rs116855232; also present in NUDT15*3) and an in-frame insertion in exon 1 (NM_018283.4:c.50_55dup, p.Gly17_Val18dup, rs746071566).

Principle of detection

The real-time PCR kit is intended for qualitative genotyping of the rs116855232 polymorphism in the human gene NUDT15 encoding the enzyme Nudix hydrolase 15. The test serves as an aid to diagnosis in patients presenting with clinical suspicion or history of adverse events caused by reduced metabolism of thiopurine drugs. The detection is performed from human genomic DNA samples by real-time PCR method (allelic discrimination).

Available products

Cat. No.	Product	rxn
3258-025	gb PHARM NUDT15 (R139C)	25
3258-050	gb PHARM NUDT15 (R139C)	50

1 kit contains reagents to provide 25 or 50 PCR reactions (20 µl volume of each reaction).

Parameters of the diagnostic kit

- ***in vitro* diagnostics**
- **CE IVD, IVDR certified**
- **ready-to-use assay**
- **sample concentration 10-100 ng/µl**
- **positive and negative controls included**
- **FAM and HEX channels detection**
- **identical amplification profile as gb PHARM, gb HEMO, gb GENETIC kits**

Content of the diagnostic kit

*	Component	Conc.	Purpose
🟢	Assay qPCR NUDT15 (R139C)	1.25×	Detection assay
🟡	Standard WT NUDT15 (R139C)	10^4 cop/µl	Positive Control
🔴	Standard MUT NUDT15 (R139C)	10^4 cop/µl	Positive Control
🟣	Standard HET NUDT15 (R139C)	10^4 cop/µl	Positive Control
⚪	Deionized Water		Negative Control

** Lid colour*



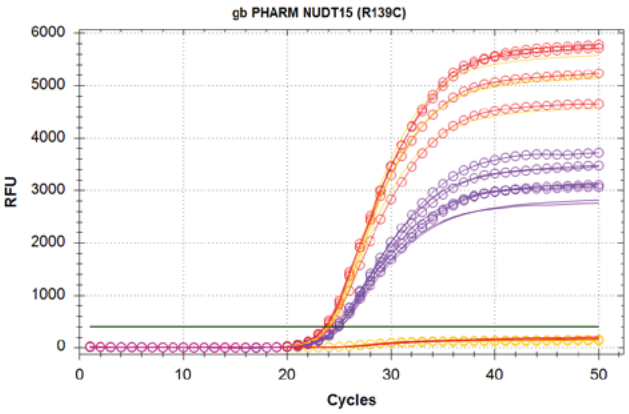


Fig. 1 – Detection of NUDT15 (R139C) standards on CFX96 device; yellow line – wild type; red line – mutant; violet line – heterozygote; smooth line – FAM channel; dotted line – HEX channel

Validated for cyclers

- CFX Opus 96 (Bio-Rad)
- CFX96/CFX96 Touch (Bio-Rad)
- LightCycler 480/Cobas z480 (Roche Diagnostics)
- QuantStudio 5 (Applied Biosystems)

CE IVD