

Human genetics

Highest precision in the detection of genetic predispositions



06/2022

**Practical:**

Same workflow and same cycler profile for all RIDA®GENE products

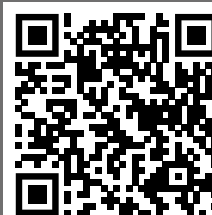
**Innovative:**

Patented detection technology for point mutations:
No melting curve analysis necessary

**Rapid:**

Results in less than 2 hours

More information:



<https://r-bio/1h>

Human genetics

Benefits



Reliable:

All controls (Human Control and Positive Control, respectively) are included in the kit



High analytical sensitivity:

- Verification of potentially interfering substances
- Precision determination



Quality:

Development and manufacturing in Germany under ISO 13485



Test format:

Kit is sufficient for 100 reactions

Human genetic diagnostics

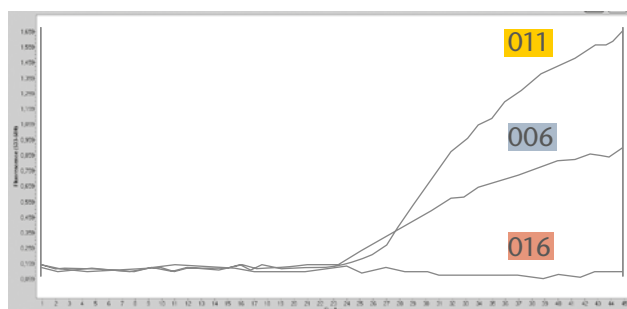
Human genetics is now of great importance in all fields of medicine, and deals with the hereditary basis of human diseases (ankylosing spondylitis, venous thromboembolism, lactose intolerance).

Our human genetics product line offers a rapid and reliable solution for the detection of genetic predispositions associated with specific diseases. This is done using special, modern SNP (single nucleotide polymorphism) technology, which detects point mutations in real-time PCR.

Early detection of genetic predispositions using the RIDA®GENE tests opens up the possibility of initiating timely therapies.

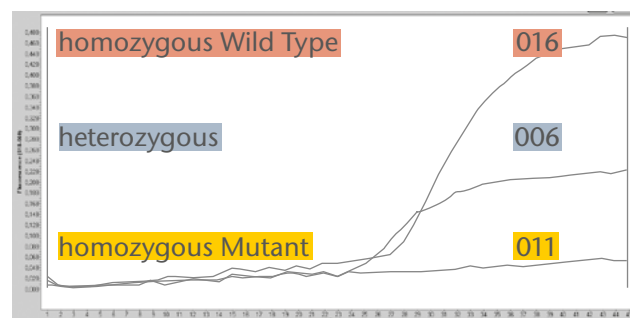
Convenient evaluation of real-time PCR using the example of RIDA®GENE Lac Intol

Detection channel 533/610



G22018 Wild type

Detection channel 618/660



22018A Mutant

Human genetics

HLA-B27

The human leukocyte antigen B27 (HLA-B27) is a major class I histocompatibility complex cell surface antigen. Its task is to present microbial antigens to T-cells.

An association with specific inflammatory, rheumatic diseases, spondyloarthritis (SpA), particularly ankylosing spondylitis (AS) is a given in carriers of HLA-B27 alleles ^(1,2). AS is a chronic, rheumatic inflammation, which mainly affects the spine and the sacroiliac joints. Other rheumatic diseases associated with HLA-B27 include Reiter's syndrome, acute anterior uveitis and inflammatory intestinal disease ⁽³⁾.

Factor II / Factor V

Thromboses are a major medical problem that increases with age ⁽⁴⁾. Factor V Leiden is considered the most common genetic factor that is described in combination with the development of venous thromboembolism ⁽⁵⁾. In this case, it is a point mutation in the factor V gene at position 1691 that leads to a base exchange from G to A ⁽⁶⁾.

Factor II, a vitamin K-dependent glycoprotein, which plays an essential role in blood coagulation, is also considered a risk factor for blood clotting disorders ^(4, 7).

The factor II variant gene can also be associated with other medical conditions like myocardial infarction or pregnancy loss ^(8,9). The susceptibility to disorders increases when multiple genetic risk factors are present (e.g., Factor V Leiden mutation, MTHFR variants, protein C deficiency, or protein S deficiency) or in the event of exposure to other risk factors, such as smoking, pregnancy, overweight, oral contraceptives, or immobility ⁽⁴⁾.

Lactose intolerance

Lactose, a disaccharide made up of galactose and glucose, is the main energy source of milk in humans and animals.

Lactose can be broken down by the enzyme lactase, thereby making the monosaccharides available for further use ⁽¹⁰⁾. Breakdown and adsorption take place in the small intestine ⁽¹¹⁾. If lactose can only be broken down in small quantities or not at all, this leads to an excessive osmotic load and increases the water content in the intestine. Furthermore, lactose gets into the large intestine, where it is fermented by intestinal bacteria, contributing to the production of short-chain fatty acids and gases such as hydrogen, carbon dioxide, and methane ⁽¹²⁾. This, in turn, can cause clinical symptoms such as bloating, abdominal pain, cramps and/or postprandial fullness, belching, diarrhea and in some cases constipation, nausea, and vomiting ^(10,12).



Information on the portfolio

Ordering information

Product	Matrix	Parameters	Art. No.
Real-time PCR			
RIDA®GENE HLA-B27	Whole blood EDTA samples	Spondylitis ankylosans (Morbus Bechterew)	PY0205
RIDA®GENE Factor II	Whole blood EDTA samples	Venous thromboembolism (G20210A)	PY1205
RIDA®GENE Factor V	Whole blood EDTA samples	Venous thromboembolism (G1691A)	PY1210
RIDA®GENE Lac Intol	Whole blood EDTA samples	Lactose intolerance (C13910T & G22018A)	PY4215



Contact us for more information:

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- ² Schlosstein L, Terasaki PI, Bluestone R, Pearson CM. High association of an HL-A antigen, W27, with ankylosing spondylitis. *N Engl J Med* 1973; 288:704-6.
- ³ Brewerton DA, Nicholls A, Caffrey M, Waters D, James DCO. HL-A 27 and arthropathies associated with ulcerative colitis and psoriasis. *Lance* 1974; 956-58.
- ⁴ Zhang S, Taylor AK, Huang X, Luo B, Spector EB, Fang P, et al. Venous thromboembolism laboratory testing (factor V Leiden and factor II c.*97G>A), 2018 update: a technical standard of the American College of Medical Genetics and Genomics (ACMG). *Genet Med*. 2018;20(12):1489-98.
- ⁵ Kujovich JL. Factor V Leiden thrombophilia. *Genet Med*. 2011;13(1):1-16.
- ⁶ Safavi-Abbasi S, Di Rocco F, Nakaji P, Feigl GC, Gharabaghi A, Samii M, et al. Thrombophilia Due to Factor V and Factor II Mutations and Formation of a Dural Arteriovenous Fistula: Case Report and Review of a Rare Entity. *Skull Base*. 2008;18(2):135-43.
- ⁷ Safavi-Abbasi S, Di Rocco F, Nakaji P, Feigl GC, Gharabaghi A, Samii M, et al. Thrombophilia Due to Factor V and Factor II Mutations and Formation of a Dural Arteriovenous Fistula: Case Report and Review of a Rare Entity. *Skull Base*. 2008;18(2):135-43.
- ⁸ Rees DC, Chapman NH, Webster MT, Guerreiro JF, Rochette J, Clegg JB. Born to clot: the European burden. *Br J Haematol*. 1999;105(2):564-6.
- ⁹ Gao H, Tao FB. Prothrombin G20210A mutation is associated with recurrent pregnancy loss: a systematic review and meta-analysis update. *Thromb Res*. 2015;135(2):339-46.
- ¹⁰ Toca MDC, Fernández A, Orsi M, Tabacco O, Vinderola G. Lactose intolerance: myths and facts. An update. *Arch Argent Pediatr*. 2022;120(1):59-66.
- ¹¹ Misselwitz B, Butter M, Verbeke K, Fox MR. Update on lactose malabsorption and intolerance: pathogenesis, diagnosis and clinical management. *Gut*. 2019;68(11):2080-91.
- ¹² Catanzaro R, Sciuto M, Marotta F. Lactose Intolerance—Old and New Knowledge on Pathophysiological Mechanisms, Diagnosis, and Treatment. *SN Comprehensive Clinical Medicine*. 2021;3:499-509.