



Patient:

BNI Lab-Nr.

<input type="checkbox"/> Familial Mediterranean Fever (FMF)	
Origin	Mediterranean basin, in particular Turkey, Armenia and Israel.
Inheritance	Recessive
Syndrome	Recurrent fevers of 1-3 (may be up to 14) days duration with peritonitis (acute abdomen), pleurisy and/or other serosities. Can be complicated by amyloidosis ultimately causing renal failure.
Test	DNA extraction, polymerase chain reaction (PCR), DNA sequencing the <i>MEFV</i> gene in the order of exon 10 (~88% of disease-associated mutations), exon 2 (11% of mutations), exons 3, 5, 1, 8, 9 and the promoter until an FMF genotype (2 pathogenic mutations) is found. On request also individual exons 1-10 or promoter separately. Batch analysis of Exons 1-3, 5, 8-10 and promoter available.
Charges	€ 198.18 for exon 10, € 72.85 for each additional exon or promoter. Batch analysis € 525,15.

<input type="checkbox"/> Hyper-IgD and Periodic Fever Syndrome (HIDS)	
Origin	Western and central Europe
Inheritance	Recessive
Syndrome	Early-onset recurrent fevers of 3-7 days duration with rash, cervical lymphadenopathy, abdominal pain, sometimes arthritis. Rarely amyloidosis. Elevated serum IgD and IgA concentrations.
Test	DNA extraction, PCR, DNA sequencing of exons 2, 3, 4, 5, 6, 7, 8, 9, 10, and 11 of the mevalonate kinase (<i>MVK</i>) gene.
Charges	€ 514.07 total (10 Sequenzierreaktionen).

<input type="checkbox"/> TNF-Receptor-Associated Periodic Syndrome (TRAPS, Hibernian fever)	
Origin	Europe
Inheritance	Dominant
Syndrome	Recurrent fevers of more than a week's duration with abdominal pain, conjunctivitis, periorbital edema and myalgia. Occasionally amyloidosis.
Test	DNA extraction, PCR, DNA sequencing of exons 2, 3, 4, 5, 6, and 7 of the <i>TNFRSF1A</i> gene.
Charges	€ 312.43 total (6 sequencing reactions).

<input type="checkbox"/> Familial Cold Urticaria (FCU), Muckle-Wells Syndrome (MWS), CINCA Syndrome	
Origin	Europe
Inheritance	Dominant
Syndrome	Fever, rash, arthritis and conjunctivitis, either following cold exposure (FCU, syn. Familial cold autoinflammatory syndrome, FCAS), or spontaneously recurring and with hearing loss (MWS). Chronic infantile neurological, cutaneous and articular (CINCA-) syndrome of chronic meningitis with rash and arthritis of newborns.
Test	DNA extraction, PCR, DNA sequencing of exons 3 and 4 of the <i>CIAS1/NALP3</i> gene.
Charges	€ 312.43 total (6 sequencing reactions).

<input type="checkbox"/> PAPA Syndrome, Familial Recurrent Arthritis (FRA)	
Origin	Europe
Inheritance	Dominant
Syndrome	Early-onset recurrent fevers with pyogenic sterile arthritis, pyoderma gangrenosum and acne (PAPA) or arthritis only (FRA).
Test	DNA extraction, PCR, DNA sequencing of exons 10 and 11 of the <i>PSTPIP1 (CD2BP1)</i> gene.
Charges	€ 198.18 total (2 sequencing reactions).

Material	2 ml EDTA blood, please send without cooling. Include billing address.
Address	Bernhard Nocht Institute for Tropical Medicine, Lab 8, PO Box 304120, D-20324 Hamburg, Germany
Laboratory	Birgit Muntau, Sonja Burwinkel, Tel.: ++49 40 42818 514, Email: burwinkel@bni-hamburg.de
Consultation	Christian Timmann, MD, ++49 40 42818 516, timmann@bni-hamburg.de Prof. Rolf Horstmann, MD, horstmann@bni-hamburg.de

Stand 11.2008