



nuuma genetics

Service Portfolio 2026

General – Genetic Counseling

STUDY	SAMPLE SOURCE	METHODOLOGY	CONSIDERATIONS	TURNAROUND TIME (BUSINESS DAYS)
Genetic Counseling (pre-test)	-	Consultation with a Physician (video consultation or in person)	The patient must send all clinical and medical documentation, as well as their family medical history, to the laboratory by email, together with the request for a genetic counseling consultation.	-
Genetic Counseling (post-test)	-	Consultation with a Physician (video consultation or in person)	Request containing the patient's identification details and the relevant test sent to the laboratory's email address.	-

General Cytogenetics – Array-CGH

STUDY	SAMPLE SOURCE	METHODOLOGY	CONSIDERATIONS	TURNAROUND TIME (BUSINESS DAYS)
Array-CGH 60K (prenatal)	Amniotic fluid (5 mL) Container: sterile syringe or sterile collection vial	array-CGH	The patient does not need to be fasting. Processing is carried out in collaboration with an external laboratory.	7
Array-CGH 60K (postnatal)	Peripheral blood — lavender-top EDTA tube, 5 mL	array-CGH	The patient does not need to be fasting. Processing is carried out in collaboration with an external laboratory.	15
Array-CGH 180K (postnatal)	Peripheral blood — lavender-top EDTA tube, 5 mL	array-CGH	The patient does not need to be fasting. Processing is carried out in collaboration with an external laboratory.	20
Array-CGH 400K (postnatal)	Peripheral blood — lavender-top EDTA tube, 5 mL	array-CGH	The patient does not need to be fasting. Processing is carried out in collaboration with an external laboratory.	30

General Cytogenetics – Karyotype

STUDY	SAMPLE SOURCE	METHODOLOGY	CONSIDERATIONS	TURNAROUND TIME (BUSINESS DAYS)
Peripheral Blood Karyotype	Peripheral blood — green-top lithium heparin tube, 3 mL (without separator gel)	G-banded karyotyping	The patient does not need to be fasting.	20
Capillary Blood Karyotype	Capillary blood — appropriate Microtainer	G-banded karyotyping	The patient does not need to be fasting.	20

General Cytogenetics – Karyotype

STUDY	SAMPLE SOURCE	METHODOLOGY	CONSIDERATIONS	TURNAROUND TIME (BUSINESS DAYS)
Amniotic Fluid Karyotype	Amniotic fluid (15–20 mL). Container: sterile syringe or sterile collection vial.	G-banded karyotyping	The patient does not need to be fasting.	15
Products of Conception Karyotype	Fetal and/or placental tissue (chorionic villi). Container: sterile wide-mouth container.	G-banded karyotyping	-	25
Chorionic Villus Karyotype	Chorionic villi. Container: sterile wide-mouth container.	G-banded karyotyping	The patient does not need to be fasting.	15-20

Prenatal Cytogenetics – Prenatal Diagnosis

STUDY	SAMPLE SOURCE	METHODOLOGY	CONSIDERATIONS	TURNAROUND TIME (BUSINESS DAYS)
QF-PCR of Amniotic Fluid	Amniotic fluid (5 mL) + Maternal buccal mucosa (sterile swab). Amniotic fluid container: sterile syringe or sterile collection vial.	QF-PCR — Detection of aneuploidies of chromosomes 13, 18, 21, X and Y + fetal sex	The patient does not need to be fasting. <i>If the fluid sample appears bloody, due to possible maternal contamination, a buccal mucosa swab will be requested from the mother.</i>	15
QF-PCR of Chorionic Villus Samples	Chorionic villus sample + Maternal buccal mucosa (sterile swab). Chorionic villus sample container: sterile wide-mouth container.	QF-PCR — Detection of aneuploidies of chromosomes 13, 18, 21, X and Y + fetal sex	The patient does not need to be fasting. <i>To assess possible maternal contamination, a buccal mucosa swab will be requested from the mother.</i>	48 - 72 h
FISH of Spermatozoa	Semen sample (entire ejaculate). Container: sterile container. Minimum volume: 500 µL. Minimum concentration: 1 million/µL.	FISH — Analysis using specific probes for disomies of sex chromosomes X and Y and chromosomes 13, 18 and 21 + diploidies	-	48 - 72 h

Prenatal Cytogenetics – Non-Invasive Prenatal Test (NIPT)

STUDY	SAMPLE SOURCE	METHODOLOGY	CONSIDERATIONS	TURNAROUND TIME (BUSINESS DAYS)
Secure Natal Basic	Maternal peripheral blood — cell-free DNA tube (provided by Nuuma Genetics), 10 mL. <i>See note on mixing with preservative.</i>	Non-Invasive Prenatal Test (NIPT) — Screening for aneuploidies of chromosomes 13, 18, 21, X and Y + fetal sex	The patient does not need to be fasting. Processing in collaboration with an external laboratory.	3-5
Secure Natal Plus	Maternal peripheral blood — cell-free DNA tube (provided by Nuuma Genetics), 10 mL. <i>See note on mixing with preservative.</i>	Non-Invasive Prenatal Test (NIPT) — Screening for aneuploidies of all chromosomes + fetal sex + microdeletions/microduplications up to 7 Mb	The patient does not need to be fasting. Processing in collaboration with an external laboratory.	3-5
Secure Natal Expanded	Maternal peripheral blood — cell-free DNA tube (provided by Nuuma Genetics), 10 mL. <i>See note on mixing with preservative.</i>	Non-Invasive Prenatal Test (NIPT) — Screening for aneuploidies of all chromosomes + fetal sex + microdeletion/microduplication syndromes up to 7 Mb + 5 well-characterized genetic syndromes	The patient does not need to be fasting. Processing in collaboration with an external laboratory. <i>Syndromes included: DiGeorge, Prader-Willi/Angelman, Cri-du-chat, Wolf-Hirschhorn, 1p36 deletion.</i>	7-9

* **Standard shipping conditions:** All samples will be shipped at room temperature unless otherwise expressly indicated (**) in the 'Considerations' column.

* **All genetic tests must be accompanied by a medical prescription, as well as the specific informed consent form, duly completed.**

The informed consent form will be obtained and retained, in accordance with current legislation, by the physician requesting the test.

* **Cell-free DNA tube (Secure Natal):** After collection, mix the blood with the preservative by shaking the tube 10 times. Place it in a biohazard bag with its corresponding box for shipping.

* **Receipt of Reports:**

Reports are sent electronically to the source of the sample and/or the prescribing specialist, encrypted with an access password.

If the patient wishes to receive a copy, they must indicate this at the time of the request, providing their contact details on the request form (national ID number, telephone number, full name) and specifying the email address to which they would like the report(s) to be sent. For data protection purposes, the reports will be sent encrypted, and a password will be required to view their contents.

The patient may also request a copy of the reports by email at info@nuumagenetics.com, indicating that they wish to receive a copy at the email address of their choice and, in all cases, attaching a photocopy of their national ID card.

** **Turnaround time starts from the time the sample is received by the laboratory in suitable condition.**

Molecular Biology – Fragment Analysis

STUDY	SAMPLE SOURCE	METHODOLOGY	CONSIDERATIONS	TURNAROUND TIME (BUSINESS DAYS)
Congenital Bilateral Absence of the Vas Deferens (CBAVD) (OMIM: 277180)	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	68 most frequent mutations + poly-T allele — CFTR gene kit	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	10
Friedreich Ataxia (OMIM: 229300)	Peripheral blood — lavender-top EDTA tube, 5 mL	PCR + Fragment analysis — FXN gene (GAA expansion)	The patient does not need to be fasting. Processing in collaboration with an external laboratory.	25
Spinocerebellar Ataxia — SCA (OMIM: 164400, 183090, 109150, 183086, 164500)	Peripheral blood — lavender-top EDTA tube, 5 mL	PCR + Fragment analysis — SCA1, SCA2, SCA3, SCA6 and SCA7 genes (triplet repeat expansion)	The patient does not need to be fasting. Processing in collaboration with an external laboratory.	25-30
Fragile X Syndrome (FRAXA) (OMIM: 300624)	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab); Amniotic fluid (5 mL, sterile syringe or sterile vial); Chorionic villi (sterile wide-mouth container); Maternal buccal mucosa (sterile swab)**	TP-PCR — FMR1 gene (CGG expansion)	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols. ** To assess possible maternal contamination of amniotic fluid or chorionic villus samples, a buccal mucosa swab will be requested from the mother.	10
Steinert Myotonic Dystrophy (OMIM: 160900)	Peripheral blood — lavender-top EDTA tube, 5 mL	PCR + TP-PCR — DMPK gene (triplet repeat expansion)	The patient does not need to be fasting. Processing in collaboration with an external laboratory.	30
Myotonic Dystrophy Type II (OMIM: 602668)	Peripheral blood — lavender-top EDTA tube, 5 mL	PCR + TP-PCR — CNBP gene (quadruplet repeat expansion)	The patient does not need to be fasting. Processing in collaboration with an external laboratory.	30
Oculopharyngeal Muscular Dystrophy (OMIM: 164300)	Peripheral blood — lavender-top EDTA tube, 5 mL	PCR + TP-PCR — DMPK gene (triplet repeat expansion)	The patient does not need to be fasting. Processing in collaboration with an external laboratory.	25
Cystic Fibrosis — 68 Mutations (OMIM: 219700)	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	68 most frequent mutations + poly-T allele — CFTR gene kit	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	10
Male Infertility	Peripheral blood — lavender-top EDTA tube, 5 mL	Multiplex PCR + Fragment analysis — Y chromosome microdeletions, AZF region (DAZ)	The patient does not need to be fasting.	15

Molecular Biology – qPCR

STUDY	SAMPLE SOURCE	METHODOLOGY	CONSIDERATIONS	TURNAROUND TIME (BUSINESS DAYS)
Celiac Disease (OMIM: 212750)	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit)	qPCR — HLA-DQ2 (HLA-DQB1*02 and HLA-DQA1*05 alleles) and HLA-DQ8	Peripheral blood: the patient does not need to be fasting. Saliva samples: follow Nuuma Genetics protocols.	10
Factor II (Prothrombin) Deficiency	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit)	qPCR — F2:c.*97G>A	Peripheral blood: the patient does not need to be fasting. Saliva samples: follow Nuuma Genetics protocols.	10
Factor V Leiden Deficiency	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit)	qPCR — F5:c.1601G>A (p.Arg534Gln)	Peripheral blood: the patient does not need to be fasting. Saliva samples: follow Nuuma Genetics protocols.	10
DPYD Genotyping	Peripheral blood — lavender-top EDTA tube, 5 mL	qPCR — Polymorphisms 1905+1G>A, c.3679T>G, c.2846A>T and c.1129-5923C>G in the DPYD gene	The patient does not need to be fasting.	48 - 72 h
Factor XII	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit)	qPCR — F12:c.-4T>C	Peripheral blood: the patient does not need to be fasting. Saliva samples: follow Nuuma Genetics protocols.	10
Hemochromatosis	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit)	qPCR — HFE gene (H63D, C282Y)*1	Peripheral blood: the patient does not need to be fasting. Saliva samples: follow Nuuma Genetics protocols. *1 The study may also include the S65C variant upon medical prescription.	10
JAK2, Point Mutation (V617F)	Peripheral blood — lavender-top EDTA tube, 5 mL	qPCR — JAK2 (V617F)	The patient does not need to be fasting.	15
MTHFR	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit)	qPCR — MTHFR:c.665C>T (p.Ala222Val)*2	Peripheral blood: the patient does not need to be fasting. Saliva samples: follow Nuuma Genetics protocols. *2 The study may also include the A1298C variant upon medical prescription.	10
Expanded Thrombophilia Panel	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit)	qPCR — FII (Prothrombin), FV Leiden, FXII, MTHFR	Peripheral blood: the patient does not need to be fasting. Saliva samples: follow Nuuma Genetics protocols.	10

Molecular Biology – MLPA

STUDY	SAMPLE SOURCE	METHODOLOGY	CONSIDERATIONS	TURNAROUND TIME (BUSINESS DAYS)
Alpha-Thalassemia (OMIM: 604131)	Peripheral blood — lavender-top EDTA tube, 5 mL	MLPA — HBA gene	The patient does not need to be fasting.	10
Spinal Muscular Atrophy	Peripheral blood — lavender-top EDTA tube, 5 mL	MLPA — SMN1, SMN2 genes	The patient does not need to be fasting.	10
Duchenne-Becker Muscular Dystrophy (OMIM: 310200)	Peripheral blood — lavender-top EDTA tube, 5 mL	MLPA — DMD gene	The patient does not need to be fasting. Processing in collaboration with an external laboratory.	15-20
Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency (OMIM: 613815)	Peripheral blood — lavender-top EDTA tube, 5 mL	Sequencing and MLPA — CYP21A2 gene	The patient does not need to be fasting. Processing in collaboration with an external laboratory.	35
Autosomal Dominant Polycystic Kidney Disease (OMIM: 601313 / 176910)	Peripheral blood — lavender-top EDTA tube, 5 mL	MLPA — PKD1, PKD2 genes	The patient does not need to be fasting. Processing in collaboration with an external laboratory.	20

Molecular Biology – PGT

STUDY	SAMPLE SOURCE	METHODOLOGY	CONSIDERATIONS	TURNAROUND TIME (BUSINESS DAYS)
PGT-A BASIC	Trophectoderm cells (approx. 6-8). Container: specific tube provided by Nuuma	NGS, Sequencing — Detection of aneuploidies of all chromosomes up to 4 Mb	SPECIAL SHIPPING CONDITION: Freeze at -20 °C after collection (if this is not possible, keep at -4 °C). Ship at -4 °C.	15
PGT-A UPGRADE	Trophectoderm cells (approx. 6-8). Container: specific tube provided by Nuuma	NGS, Sequencing — Detection of aneuploidies of all chromosomes, heteroploidy + 42 microduplication/microdeletion syndromes up to 4 Mb	SPECIAL SHIPPING CONDITION: Freeze at -20 °C after collection (if this is not possible, keep at -4 °C). Ship at -4 °C.	15
PGT-A COMPLETE	Trophectoderm cells (approx. 6-8). Container: specific tube provided by Nuuma + parental sample: peripheral blood — lavender-top EDTA tube, 5 mL; saliva (Nuuma saliva kit) or buccal mucosa (sterile swab)	NGS, Sequencing — Detection of aneuploidies of all chromosomes, heteroploidy + 42 microduplication/microdeletion syndromes + parental origin of aneuploidy up to 4 Mb	SPECIAL SHIPPING CONDITION: Freeze at -20 °C after collection (if this is not possible, keep at -4 °C). Ship at -4 °C.	25
PGT-SR	Trophectoderm cells (approx. 6-8). Container: specific tube provided by Nuuma	NGS, Sequencing — Detection of unbalanced structural rearrangements and aneuploidies up to 1 Mb + complementary tests	SPECIAL SHIPPING CONDITION: Freeze at -20 °C after collection (if this is not possible, keep at -4 °C). Ship at -4 °C.	15

Molecular Biology – Sequencing (Sanger, NGS)

STUDY	SAMPLE SOURCE	METHODOLOGY	CONSIDERATIONS	TURNAROUND TIME (BUSINESS DAYS)
Achondroplasia; Thanatophoric Dysplasia Types I and II (OMIM: 100800; 187600; 187601)	Prenatal: Amniotic fluid (sterile syringe or sterile collection vial); Chorionic villi (sterile wide-mouth container). Postnatal: Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	Sanger sequencing — FGFR3 gene	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	10 (prenatal) 15 (postnatal)
Alport Syndrome (OMIM: 301050)	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — COL4A3, COL4A4, COL4A5 genes	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	30
Alzheimer's Disease Susceptibility	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	Sanger sequencing — APOE (alleles ε2 [Cys130, Cys176], ε3 [Cys130, Arg176] and ε4 [Arg130, Arg176])	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	15
BRCA1/BRCA2, Complete Sequencing	Sangre Periférica — Tubo EDTA tapón lila, 5 mL; Saliva (kit saliva Nuuma); Mucosa bucal (torunda estéril)	NGS, Sequencing — BRCA1, BRCA2 genes	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	30
CADASIL — Cerebral Autosomal Dominant Arteriopathy with Subcortical Infarcts and Leukoencephalopathy (OMIM: 125310)	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — NOTCH3 gene	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	30
Familial Cancer	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — Associated gene panel	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	30
Hereditary Breast/Ovarian Cancer — HBOC	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — Associated gene panel	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	30
Charcot-Marie-Tooth Disease	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — Associated gene panel	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	30
Diamine Oxidase (DAO) Deficiency	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	Sanger sequencing — Most frequent polymorphisms in the AOC1 gene (rs10156191, rs1049742, rs1049793 and rs2052129)	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	15
MODY Diabetes (OMIM: 125850; 125851; 600496; 606392)	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — Associated gene panel	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	30
Macular and Retinal Dystrophies	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — Associated gene panel	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols. Processing in collaboration with an external laboratory.	40
Skeletal Dysplasias	Prenatal: Amniotic fluid (sterile syringe or sterile collection vial); Chorionic villi (sterile wide-mouth container). Postnatal: Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — Associated gene panel	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols. Processing in collaboration with an external laboratory.	20 (prenatal) 30 (postnatal)

Molecular Biology – Sequencing (Sanger, NGS)

STUDY	SAMPLE SOURCE	METHODOLOGY	CONSIDERATIONS	TURNAROUND TIME (BUSINESS DAYS)
Ehlers-Danlos Syndrome	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — Associated gene panel	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols. Processing in collaboration with an external laboratory.	30
Parkinson's Disease	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — Associated gene panel	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols. Processing in collaboration with an external laboratory.	35
Epilepsy	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — Associated gene panel	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols. Processing in collaboration with an external laboratory.	35
Cystic Fibrosis — Complete Sequencing (OMIM: 219700)	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Complete sequencing — CFTR gene	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	30
Familial Mediterranean Fever (OMIM: 608107)	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	MEFV gene	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	30
JAK2, Exon 12	Peripheral blood — lavender-top EDTA tube, 5 mL;	Sanger sequencing — JAK2 (exon 12)	The patient does not need to be fasting. Processing in collaboration with an external laboratory.	20
Laminopathy (LMNA)	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — LMNA gene	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	30
Lynch Syndrome	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — Associated gene panel	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	30
Marfan Syndrome (OMIM: 154700)	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — Associated gene panel	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols. Processing in collaboration with an external laboratory.	30
Genetic Matching	-	NGS, Sequencing — Custom gene panel (report)	-	7
Familial Transthyretin Amyloid Cardiomyopathy (OMIM: 105210)	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	Sanger sequencing — TTR gene	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	30
Familial Transthyretin Amyloid Cardiomyopathy	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — Associated gene panel	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	30

Molecular Biology – Sequencing (Sanger, NGS)

STUDY	SAMPLE SOURCE	METHODOLOGY	CONSIDERATIONS	TURNAROUND TIME (BUSINESS DAYS)
Dilated Cardiomyopathy	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — Associated gene panel	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	30
Hypertrophic Cardiomyopathy	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Complete sequencing — CFTR gene	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	30
Left Ventricular Noncompaction Cardiomyopathy	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Complete sequencing — CFTR gene	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	30
Point Mutation	Peripheral blood — lavender-top EDTA tube, 5 mL;	Sanger sequencing — Single-variant analysis	Peripheral blood: the patient does not need to be fasting.	15
Neurofibromatosis Types I and II	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — NF1, NF2 genes	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols. Processing in collaboration with an external laboratory.	30
Leber Hereditary Optic Neuropathy	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — Associated gene panel*	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols. Processing in collaboration with an external laboratory. * May include mitochondrial complex MTND4, MTND6 and MTND1 (mutations m.11778G>A, m.14484T>C, m.3460G>A).	35
Noonan Syndrome (RASopathies)	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — Associated gene panel	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols. Processing in collaboration with an external laboratory.	30
Familial Adenomatous Polyposis	-	NGS, Sequencing — Custom gene panel (report)	-	7
Secure Match Basic (incl. MLPA HBA1 and HBA2, SMN1 and FRAXA screening)	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	Sanger sequencing — TTR gene	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	30
Secure Match Expanded (incl. MLPA HBA1 and HBA2, SMN1 and FRAXA screening)	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — Associated gene panel	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	30
Brugada Syndrome (OMIM: 601144)	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit); Buccal mucosa (sterile swab)	NGS, Sequencing — Associated gene panel	Peripheral blood: the patient does not need to be fasting. Saliva or buccal mucosa samples: follow Nuuma Genetics protocols.	30

Molecular Biology – Other

STUDY	SAMPLE SOURCE	METHODOLOGY	CONSIDERATIONS	TURNAROUND TIME (BUSINESS DAYS)
HLA-C	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit)	PCR-SSP — Determination of HLA-C c1 and/or C2 haplotype	Peripheral blood: the patient does not need to be fasting. Saliva samples: follow Nuuma Genetics protocols.	10
HPV	Cervical swab; Paraffin-embedded tissue; Liquid-based cytology. Container: according to sample type (liquid-based cytology vial, paraffin cassette or swab in transport medium)	PCR-SSOP — HPV genotyping	-	15
KIR	Peripheral blood — lavender-top EDTA tube, 5 mL; Saliva (Nuuma saliva kit)	PCR-SSP — Genes 2DL1, 2DL2, 2DL3, 2DL4, 2DL5, 2DS1, 2DS2, 2DS3, 2DS4, 2DS5, 3DL1, 3DL2, 3DL3, 3DS1, 2DP1, 3DP1	Peripheral blood: the patient does not need to be fasting. Saliva samples: follow Nuuma Genetics protocols.	10

* **Standard shipping conditions:** All samples will be shipped at room temperature unless otherwise expressly indicated (**) in the 'Considerations' column.

* **PGT-A / PGT-SR:** special condition — freeze at -20 °C after collection and ship at -4 °C.

* **Saliva samples:** follow the specifications in the sample collection protocol provided by Nuuma Genetics.

* **All genetic tests must be accompanied by a medical prescription and the specific informed consent form, duly completed.**
The informed consent form will be obtained and retained, in accordance with current legislation, by the physician requesting the test.

* **Receipt of Reports.**
Reports are sent electronically to the source of the sample and/or the prescribing specialist, encrypted with an access password.
If the patient wishes to receive a copy, they must indicate this at the time of the request, providing their contact details on the request form (national ID number, telephone number, full name) and specifying the email address to which they would like the report(s) to be sent. For data protection purposes, the reports will be sent encrypted, and a password will be required to view their contents.
The patient may also request a copy of the reports by email at info@nuumagenetics.com, indicating that they wish to receive a copy at the email address of their choice and, in all cases, attaching a photocopy of their national ID card.

** **Turnaround time starts from the time the sample is received by the laboratory in suitable condition.**

Exomes and WGS

STUDY	SAMPLE SOURCE	METHODOLOGY	CONSIDERATIONS	TURNAROUND TIME (BUSINESS DAYS)
Targeted Exome < 50 Genes	Prenatal: Amniotic fluid (sterile syringe or sterile collection vial). Postnatal: Peripheral blood — lavender-top EDTA tube, 5 mL.	NGS, Sequencing — Associated gene panel	The patient does not need to be fasting.	30
Targeted Exome > 50 Genes	Prenatal: Amniotic fluid (sterile syringe or sterile collection vial). Postnatal: Peripheral blood — lavender-top EDTA tube, 5 mL.	NGS, Sequencing — Associated gene panel	The patient does not need to be fasting.	40
WES EXTRA	Peripheral blood — lavender-top EDTA tube, 5 mL.	NGS, Sequencing — Filtering based on HPO criteria and/or custom panel	The patient does not need to be fasting. Processing in collaboration with an external laboratory.	40
Trio Exome	Peripheral blood — lavender-top EDTA tube, 5 mL.	NGS, Sequencing — Filtering based on HPO criteria and/or custom panel	The patient does not need to be fasting. Processing in collaboration with an external laboratory.	30 (prenatal) 40 (postnatal)

* **Standard shipping conditions:** All samples will be shipped at room temperature.

* **All genetic tests must be accompanied by a medical prescription and the specific informed consent form, duly completed.**
The informed consent form will be obtained and retained, in accordance with current legislation, by the physician requesting the test.

* **Receipt of Reports.**

Reports are sent electronically to the source of the sample and/or the prescribing specialist, encrypted with an access password.

If the patient wishes to receive a copy, they must indicate this at the time of the request, providing their contact details on the request form (national ID number, telephone number, full name) and specifying the email address to which they would like the report(s) to be sent. For data protection purposes, the reports will be sent encrypted, and a password will be required to view their contents.

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** **Turnaround time starts from the time the sample is received by the laboratory in suitable condition.**



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