healthincode
> Neurology

+34 881 60 00 03
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STUDY REQUISITION FORM

1 Patient and sample information

Patient
First and last name

Date of birth
DD/MM/YY

Sex ○ Female ○ Male

Sample collection date

☐ Blood
Peripheral blood from 3 to 5 ml in EDTA tubes

☐ Saliva
Using the indicated saliva kit

Sample reference: use the same reference on the collection tube

☐ DNA*
Minimum 5 µg and concentration 50 ng/mL for DNA-derived from blood, saliva, tissue (fresh or frozen).
Minimum10 µg and concentration 50 ng/mL for DNA-derived from paraffin-embedded tissue.
***DNA source:**
Blood, frozen blood, saliva, fresh tissue, frozen tissue, paraffin-embedded tissue, etc.

2 Information of the requesting physician

First and last name

Hospital/Institution

Address | City

Province / Region / State | Country | Zip code

Phone | Email

3 Authorized person(s) to receive the results

First and last name

E-mail
to receive results

First and last name

E-mail
to receive results

In compliance with the Spanish and European personal data protection laws, the results will only be delivered to the persons duly identified in this requisition form.

4 Invoicing details

☐ **Hospital / Institution**

☐ **Self-pay patient**
Payment method: ○ Bank transfer ○ Credit card

Name of the hospital or patient
name that should appear on the invoice

National ID / Tax number

Address | City

Province / Region / State | Country | Zip code

Phone | E-mail
to send the invoice

Contact person

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5 Genetic study requested

Sequencing Panels

Genetic Muscle Disorders [GMD]

- ☐ S-202008553 Congenital structural GMD [78 genes]
- ☐ S-202008554 Child- and adult-onset structural GMD [64 genes]
- ☐ S-202008552 Limb-girdle muscular dystrophies [43 genes]
- ☐ S-202008620 Distal myopathies [37 genes]
- ☐ S-202008621 Myofibrillar myopathies with protein aggregates [20 genes]
- ☐ S-201602251 Emery-Dreifuss-type muscular dystrophies [7 genes]
- ☐ S-201602252 Dystrophinopathies [DMD gene] NGS sequencing
- ☐ S-202008623 Myopathies related to glycogen metabolism [21 genes]
- ☐ S-201804629 Myopathies related to lipid metabolism [15 genes]
- ☐ S-202008622 Nuclear mitochondrial myopathies [69 genes]
- ☐ S-202008642 Rhabdomyolysis and hyperCKemia [47 genes]
- ☐ S-202008624 Non-dystrophic myotonies [10 genes]
- ☐ S-202008629 Congenital myasthenia extended panel [29 genes]
- ☐ S-202008634 Congenital myasthenia basic panel [6 genes]
- ☐ S-202008626 Arthrogryposis extended panel [86 genes]
- ☐ S-202008665 Multiple pterygium/Escobar and related syndromes [15 genes]
- ☐ S-202008527 Distal arthrogryposis [11 genes]
- ☐ S-202008646 Structural genetic disorders comprehensive panel [133 genes]
- ☐ S-202008656 Metabolic myopathies comprehensive panel [109 genes]
- ☐ S-202008374 GMD comprehensive panel [330 genes]

Hereditary Neuropathies

- ☐ S-202008627 Charcot-Marie-Tooth disease extended panel [77 genes]
- ☐ S-202008637 Demyelinating/intermediate CMT panel [37 genes]
- ☐ S-202008636 Axonal/Intermediate CMT panel [57 genes]
- ☐ S-202008630 CMT basic panel [4 genes]
- ☐ S-202008639 Motor neuropathy/SMN1-negative spinal muscular atrophy panel [38 genes]
- ☐ S-202008641 Hereditary sensory and autonomic neuropathy panel [28 genes]
- ☐ S-202008638 Metabolic neuropathy panel [24 genes]
- ☐ S-202008640 Optic neuropathy panel [13 genes]
- ☐ S-202008657 Neuropathies comprehensive panel [150 genes]

Hereditary Spastic Paraplegia

- ☐ S-202008662 Pure spastic paraplegia [36 genes]
- ☐ S-202008661 Complex spastic paraplegia [90 genes]
- ☐ S-202008635 Spastic paraplegia basic panel [8 genes]
- ☐ S-202008658 Spastic paraplegia comprehensive panel [107 genes]

Ataxia

- ☐ S-202008530 Spinocerebellar ataxia [84 genes]
- ☐ S-202008531 Autosomal dominant spinocerebellar ataxia [24 genes]
- ☐ S-202008532 Autosomal recessive spinocerebellar ataxia [65 genes]
- ☐ S-202008529 Spastic ataxia and ataxia-dystonia syndromes [35 genes]
- ☐ S-202008528 Episodic ataxia [8 genes]
- ☐ S-202008533 Ataxia and atrophy/pontocerebellar hypoplasia [31 genes]
- ☐ S-202008375 Ataxia comprehensive panel [262 genes]

Dementia

- ☐ S-202008557 Alzheimer's disease [5 genes]
- ☐ S-202008631 Frontotemporal dementia basic panel [11 genes]
- ☐ S-202008628 Frontotemporal dementia extended panel [22 genes]
- ☐ S-202008565 Amyotrophic lateral sclerosis - Frontotemporal dementia [15 genes]
- ☐ S-202008644 Dementia comprehensive panel [49 genes]

Amyotrophic Lateral Sclerosis [ALS]

- ☐ S-202008633 Amyotrophic lateral sclerosis basic panel [3 genes]
- ☐ S-202008565 Amyotrophic lateral sclerosis - Frontotemporal dementia [15 genes]
- ☐ S-202008651 Amyotrophic lateral sclerosis and primary lateral sclerosis comprehensive panel [38 genes]

Movement Disorders

- ☐ S-202008645 Dystonia comprehensive panel [48 genes]
- ☐ S-202008546 Isolated dystonia [8 genes]
- ☐ S-202008547 Myoclonic dystonia [2 genes]
- ☐ S-202008550 Dystonia-parkinsonism [5 genes]
- ☐ S-202008548 Paroxysmal dystonia with other dyskinesias [4 genes]
- ☐ S-202008559 Parkinson's disease and related disorders [25 genes]
- ☐ S-202008632 Parkinson's disease basic panel [8 genes]
- ☐ S-202008663 Young-onset parkinsonism [8 genes]
- ☐ S-201805729 Chorea and Huntington-like disorders [19 genes]
- ☐ S-202008643 Basal ganglia calcification comprehensive panel [13 genes]
- ☐ S-201805369 Aicardi-Goutières syndrome [7 genes]
- ☐ S-202008625 Neurodegeneration with brain iron accumulation syndromes (NBIAS) [14 genes]
- ☐ S-201804729 Paroxysmal movement disorders [18 genes]
- ☐ S-202008276 Neuronal ceroid lipofuscinosis [11 genes]
- ☐ S-202008660 Metabolic movement disorders comprehensive panel [32 genes]
- ☐ S-202008659 Movement disorders comprehensive panel [152 genes]

Leukodystrophies and Other Hereditary Leukoencephalopathies

- ☐ S-202008607 POLR3-related leukodystrophy [5 genes]
- ☐ S-202008560 Pelizaeus-Merzbacher disease (PMD) and PMD-like diseases (PMLD) [5 genes]
- ☐ S-202008679 Tricotiodystrophy/Tay syndrome [5 genes]
- ☐ S-202008613 Leukodystrophies with intracranial calcifications [24 genes]
- ☐ S-202008615 Leukodystrophies with white matter rarefaction or cystic lesions on MRI [29 genes]
- ☐ S-202008667 Childhood ataxia with central nervous system hypomyelination/vanishing white matter [CACH/VWM] [5 genes]
- ☐ S-202008605 Megalencephalic leukoencephalopathy with subcortical cysts [2 genes]
- ☐ S-202008612 Leukodystrophies with spinal cord involvement on MRI [5 genes]
- ☐ S-202008614 Leukodystrophies with abnormal peaks on magnetic resonance spectroscopy [4 genes]
- ☐ S-202008606 Metachromatic leukodystrophy [3 genes]
- ☐ S-202008610 Leukodystrophies associated with lysosomal disorders [22 genes]
- ☐ S-202008611 Leukodystrophies associated with peroxisomal disorders [19 genes]

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- ☐ S-202008608 Leukodystrophies associated with energy and mitochondrial metabolism disorders [16 genes]
- ☐ S-202008609 Leukodystrophies associated with inborn errors of intermediate metabolism [17 genes]
- ☐ S-202008616 Vascular leukoencephalopathies [12 genes]
- ☐ S-202008654 Hypomyelinating leukodystrophies comprehensive panel [40 genes]
- ☐ S-202008653 Leukodystrophies due to inborn errors of metabolism comprehensive panel [73 genes]
- ☐ S-202008655 Leukodystrophies and other hereditary leukoencephalopathies comprehensive panel [142 genes]

Cerebrovascular Diseases

- ☐ S-202008524 Stroke and migraine [5 genes]
- ☐ S-202008536 Cerebral cavernoma [3 genes]
- ☐ S-201906329 CADASIL [the NOTCH3 gene]
- ☐ S-202008556 Cerebral microangiopathy [8 genes]
- ☐ S-202008558 Moyamoya disease [8 genes]
- ☐ S-202008648 Cerebrovascular diseases comprehensive panel [36 genes]

Mitochondrial diseases

- ☐ S-202008538 Specific mitochondrial respiratory chain complexes/OXPHOS deficiency panel [94 genes]
- ☐ S-202008540 mtDNA depletion [18 genes]
- ☐ S-202008674 Leigh syndrome caused by nuclear DNA mutations [70 genes]
- ☐ S-201906357 Pyruvate dehydrogenase (PDH) deficiency [12 genes]
- ☐ S-202008539 Primary coenzyme Q deficiency [13 genes]
- ☐ S-202008652 Mitochondrial nuclear genes comprehensive panel [400 genes]
- ☐ S-201805389 Mitochondrial genome [37 genes]
- ☐ S-201805390 Mitochondrial genome associated with another NGS panel

Epilepsy

- ☐ S-202008555 Neonatal and early-onset epileptic encephalopathy [90 genes]
- ☐ S-202008671 Dravet syndrome and febrile convulsions plus [16 genes]
- ☐ S-202008676 Rett and Rett-like syndrome [41 genes]
- ☐ S-202008666 Angelman-like syndrome panel [12 genes]
- ☐ S-201907249 Tuberous sclerosis [2 genes]
- ☐ S-202008561 Childhood absence epilepsy [5 genes]
- ☐ S-202008562 Focal epilepsy and other forms of familial epilepsy [33 genes]
- ☐ S-202008650 Myoclonic epilepsy comprehensive panel [43 genes]
- ☐ S-202008563 Juvenile myoclonic epilepsy [7 genes]
- ☐ S-202008564 Progressive myoclonic epilepsy [36 genes]
- ☐ S-202008602 Hyperekplexia and paroxysmal disorders related to epilepsy [9 genes]
- ☐ S-202008647 Epileptic encephalopathy comprehensive panel [124 genes]
- ☐ S-202008649 Epilepsy comprehensive panel [271 genes]

CNS Anomalies

- ☐ S-202008537 Neural tube closure defects [5 genes]
- ☐ S-202008526 Brain midline/regionalization alterations [121 genes]
- ☐ S-202008604 Holoprosencephaly [14 genes]
- ☐ S-202008566 Schizencephaly [4 genes]
- ☐ S-202008525 Agenesis of the corpus callosum [106 genes]
- ☐ S-202008681 Neuronal migration disorders/cortical dysplasias [57 genes]
- ☐ S-202008617 Lissencephaly [25 genes]
- ☐ S-202008601 Periventricular nodular heterotopia [9 genes]
- ☐ S-202008567 Band heterotopia [the DCX gene]
- ☐ S-202008664 Polymicrogyria [24 genes]
- ☐ S-202008618 Megalencephaly-polymicrogyria and dysplastic megalencephaly [7 genes]
- ☐ S-202008619 Microcephaly [88 genes]
- ☐ S-202008603 Pontocerebellar hypoplasia [18 genes]

Neurodevelopmental Disorders and Related Genetic Syndromes

- ☐ S-202008543 Intellectual disability and/or autism [865 genes]
- ☐ S-202008542 Intellectual disability [798 genes]
- ☐ S-202008534 Autism [196 genes]
- ☐ S-202008544 Intellectual disability and/or autism and epilepsy [117 genes]
- ☐ S-202008535 BAFopathies (Coffin-Siris syndrome and Nicolaides-Baraitser syndrome) [10 genes]
- ☐ S-201907249 Tuberous sclerosis [2 genes]
- ☐ S-201805369 Aicardi-Goutières syndrome [7 genes]
- ☐ S-202008669 Cockayne syndrome [5 genes]
- ☐ S-202008670 Cornelia de Lange syndrome [10 genes]
- ☐ S-202008672 Joubert syndrome [34 genes]
- ☐ S-202008673 Kabuki syndrome [2 genes]
- ☐ S-202008675 Meckel syndrome [13 genes]
- ☐ S-201906395 RASopathies syndromes [26 genes]
- ☐ S-202008677 Rubinstein-Taybi syndrome [2 genes]
- ☐ S-202008678 Seckel syndrome [9 genes]
- ☐ S-202008680 Sotos syndrome [3 genes]

Other tests:

- ☐ S-202008723 FXS/FXTAS/FXPOI [FMR1 expansions]
- ☐ S-202009944 Prader-Willi/Angelman syndromes [MS-MLPA of the PWS/AS genomic region]
- ☐ S-202008666 Angelman-like syndrome panel [12 genes]
- ☐ S-202009939 Rett syndrome [Sanger sequencing of the MECP2 gene]
- ☐ S-202009941 Rett syndrome [gene dosage analysis of the MECP2 gene by MLPA]
- ☐ S-202008676 Rett and Rett-like syndrome [41 genes]
- ☐ S-202009943 Beckwith-Wiedemann/Silver-Russell syndromes [MS-MLPA region 11p15]
- ☐ S-202009942 Silver-Russell syndrome [MS-MLPA chromosome 7]

- ☐ S-202008668 Beckwith-Wiedemann-like syndrome [8 genes]
- ☐ S-202009940 CHARGE syndrome [sequencing of the CHD7 gene]
- ☐ S-202009388 KBG syndrome [Sanger sequencing of the ANKRD11 gene]

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Nucleotide expansions

- ☐ S-202008703 Oculopharyngeal muscular dystrophy [*PABPN1* expansions]
- ☐ S-201804669 Myotonic dystrophy type 1 [*DMPK* expansions]
- ☐ S-202008721 Myotonic dystrophy type 2 [*CNBP* expansions]
- ☐ S-201601805 Friedreich ataxia [*FXN* expansions]
- ☐ S-202008193 SCA expansions - Panel 1 [SCA1, SCA2, SCA3, SCA6, SCA7]
- ☐ S-202008722 SCA expansions - Panel 2 [SCA10, SCA12, SCA17]
- ☐ S-202008724 DRPLA [*ATN1* expansions]
- ☐ S-202008723 FXS/FXTAS/FXPOI [*FMR1* expansions]
- ☐ S-201805509 C9orf72-related ALS/FTD [*C9orf72* expansions]
- ☐ S-202008725 Kennedy disease [*AR* expansions]
- ☐ S-202008726 Huntington's disease [*HTT* expansions]
- ☐ S-202008727 Huntington disease-like type 2 [*JPH3* expansions]
- ☐ S-202008728 Unverricht–Lundborg disease [*CSTB* expansions]

MLPA

- ☐ S-201602259 Dystrophinopathy [DMD gene dosage by MLPA]
- ☐ S-201703888 CMT1A/HNPP [PMP22 gene dosage by MLPA]
- ☐ S-201906211 Spinal muscular atrophy [SMN1-SMN2 gene dosage by MLPA]

Other genetic tests

- ☐ S-202109974 Individual sequencing of genes (Sanger)
- ☐ S-202109975 NextGenDx® massive sequencing
- ☐ S-202109976 Massive sequencing with CNVs

Whole exome:

- ☐ S-202110014 Whole-exome - sequencing only (fastq)
- ☐ S-202110013 Whole-exome - annotation of variants
- ☐ S-202110336 Whole-exome - with report tool
- ☐ S-202110015 Whole-exome - with clinical report
- ☐ S-202109977 Targeted exome
  Gene/genes:
- ☐ S-202110133 Trio clinical exome
- ☐ S-202109983 MLPA and methylation-specific MLPA:
  Gene/genes:

SNP array:

- ☐ S-201601485 Index case
- ☐ S-201702726 Family study or confirmation of CNVs

Array CGH:

- ☐ S-202008036 Prenatal array (37K)
- ☐ S-202109987 Postnatal array (60K)
- ☐ S-202109988 Postnatal array (180K)
- ☐ S-202109998 Variant segregation/Family studies
  Variant:
- ☐ Other services:

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6 Clinical data

We recommend attaching a clinical report to ensure the correct interpretation of the findings

7 Statement of the existence of informed consent

- ☐ The patient identified in this requisition (or his/her legal representative) is aware of the information included in it and authorizes that his/her sample be submitted for genetic testing and that a report is issued with the corresponding results.
- ☐ It is possible to obtain unexpected information during the sample analysis process, which the patient identified in this requisition (or his/her legal representative) has agreed to be informed about.
- ☐ In addition, the patient identified in this requisition (or his/her legal representative) authorizes that his/her biological sample be stored for subsequent studies and/or confirmation tests.
- ☐ The patient identified in this requisition (or his/her legal representative) also authorizes that his/her biological sample be used for research purposes approved by the relevant ethical committee, always maintaining the patient's anonymity.

Physician's signature

Date

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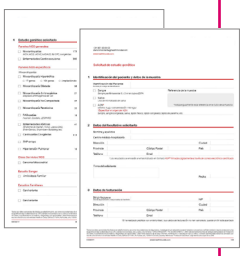
8 Sample requirements and shipping



STUDY REQUISITION

The sample for genetic testing must be sent together with a correctly filled requisition form.

Available at healthincode.com or by request at customercare@healthincode.com

SAMPLE COLLECTION

Peripheral blood*

3 to 5 ml in EDTA tubes

Genomic DNA*

NGS > 5-10 µg (A260/280 = 1.8-1.9)
Sanger > 1 µg (A260/280 = 1.8-1.9)

Saliva sample

Please use the indicated kit for sample collection.

You can request it at customercare@healthincode.com

**For delivery in over 48 h, controlled-temperature shipment (4-8 ºC) is recommended*

SAMPLE PACKAGING

Each primary container (sample tube**) must be placed inside a secondary container (sealed plastic bag or Falcon tube) with enough absorbent material. Secondary recipients must be secured inside a rigid package or box with appropriate cushioning material.

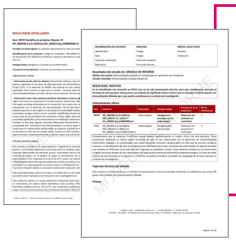
** Please make sure that the sample tube is labeled with the patient's details or reference.

SAMPLE SHIPMENT

Schedule your shipment so that sample reception takes place Monday to Thursday between 8:00 and 17:00.

HEALTH IN CODE S. L.
Edificio O Fortín, As Xubias s/n. Campus de Oza. 15006 A Coruña, Spain
Tel: +34 881 600 003

If you wish, you can request our sample pick-up service at customercare@healthincode.com



RESULTS

We will deliver our report via:

- Certified email
- Health in Code Client Portal

OUR STUDIES ALWAYS INCLUDE THE POSSIBILITY OF PRE-TEST AND POST-TEST COUNSELLING

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