NIMGenetics
New Integrated Medical Genetics

Datos Personales del Paciente

Name and Surname	Gender	Age

CGH ARRAYS

- ☐ KaryoNIM® Postnatal 60K (CGH1001)
- ☐ KaryoNIM® Postnatal 180K (CGH1003)
- ☐ KaryoNIM® 180K Autism (CGH1005)
- ☐ KaryoNIM® 400K (CGH1007)

PANELES NGS Y GENES ÚNICOS

- ☐ NIMSeq® (Reference*): _____
  Disease and/or genes: _____

EXONIC SEQUENCING

- ☐ ExoNIM® Targeted (Reference*): _____
  Disease and/or genes: _____
- ☐ ExoNIM® Extensions (Reference*): _____
  Previous study identifier: _____
  Disease and/or genes: _____
- ☐ Combination of ExoNIM® panels (Referencia*): _____
  Disease and/or genes: _____
- ☐ ExoNIM® Clínico (EXN2003)
- ☐ ExoNIM® Trío (EXN3001)
- ☐ ExoNIM® Plus Epilepsy (EXN5001)
- ☐ ExoNIM® Personalized (Reference or attach gene list): _____

OTHERS

- ☐ MLPA ((Specify/Reference*) _____
- ☐ Mitochondrial DNA ((Specify/Reference*) _____
- ☐ Triplet Expansion ((Specify/Reference*) _____

STUDY OF CARRIERS (SANGER)

- Degree of kinship with the reference case:

- Reference case previously studied at NIMGenetics:

  ☐ NO → IT IS NECESSARY TO ATTACH A COPY OF THE PREVIOUS REPORT containing information on the variants to be analysed (gene #NM, nucleotide variation/protein)

  ☐ Yes → Reference case identifier: _____ :

Variant 1: _____ (Gene) _____ (Nucleotide variation/Protein)

Variant 2: _____ (Gene) _____ (Nucleotide variation/Protein)

Variant 3: _____ (Gene) _____ (Nucleotide variation/Protein)

(*) Check our extensive portfolio for postnatal diagnosis at www.nimgenetics.com

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POSTNATAL DIAGNOSIS

SERVICE REQUEST FORM

Petitioner's Data

Requesting Entity		Date
Name and Surname		E-mail address
Address		
Province	Postal code	Telephone number

Patient's Clinical Data

Name and Surname	Gender	Age

Patient's Ethnicity

☐ Caucasian ☐ Hispanic ☐ African
☐ Asian ☐ Others _____

IMPORTANT

For proper sample processing and analysis of results, indicate the suspected diagnosis, reason for consultation, relevant clinical data and personal and family background associated with the patient, in the field provided on page 3 of this form.

Type of Sample	Date of extraction

The sample must always be accompanied by this request form and the corresponding informed consent, which can be downloaded at www.nimgenetics.com. Only if the informed consent is not enclosed with the sample, check the following box:

☐ **I declare that the patient has been informed about the indication, purpose, characteristics, scope and limitations of the requested study. The informed consent associated with this test has been signed by the patient, and it remains under the custody of the centre Hospital or medical practitioner in charge**

Signature of the medical practitioner in charge _____

Invoicing and Payment Information

Entity	
Address	
Authorised Person	Tax ID (NIF/CIF)
Telephone number	E-mail address
Telephone number	

Contact Details

NIMGenetics
Calle Faraday, 7
28049 Madrid
Phone. (+34) 91 037 83 54
contactoespecialistas@nimgenetics.com

Data Protection and Confidentiality

In accordance with Law 41/2002 Regulating Patient Autonomy and Law 15/1999 on Personal Data Protection, the petitioner shall have the consent of the patient to carry out the diagnostic tests requested and for the processing of their data. Thus, and as information to be provided to the patient, we must inform you that the data collected in this form shall be incorporated into a confidential automated file, duly registered with the Spanish Data Protection Agency, in accordance with the terms established in Law 15/1999, the ownership of which corresponds to NIMGenetics, SL, with the purpose of the processing diagnostic study described in the form, the patient being able to exercise at any time the rights to access, rectification, cancellation or objection, recognised by the regulations cited relating to Personal Data Protection, to be sent to the following address: NIMGENETICS, S.L., Genómica y Medicina, C/ Faraday, 7 28049 Madrid.

NIMGenetics is a Genetic Diagnosis centre authorised by the Department of Health and Consumption of the Community of Madrid, registered in the corresponding Register under number CS 10673

NIMGenetics S.L. - Parque científico de Madrid - C/Faraday, 7 (Campus de Cantoblanco) 28049 Madrid - CIF B-85332138

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Patient's Personal Data

Name and Surname	Gender	Age
Diagnosis		
Reason for consultation / Summary of Medical History / Background		

Clinical features (1)

Malformations - Cerebral/Abnormal Images

- ☐ Basal ganglia abnormalities
- ☐ Agenesis of the corpus callosum
- ☐ Cortical dysplasia
- ☐ Hemimegalencephaly
- ☐ Heterotopia
- ☐ Holoprosencephaly
- ☐ Hydrocephalus
- ☐ Lisencephaly
- ☐ Macrocephaly
- ☐ Microcephaly
- ☐ Periventricular leukomalacia
- ☐ Other _____

Malformations - Skeletal or Other

- ☐ Vertebral abnormalities
- ☐ Limb abnormalities
- ☐ Muscle contractions
- ☐ Craniosynostosis
- ☐ Dysmorphic features
- ☐ Scoliosis
- ☐ Fractures
- ☐ Cleft lip/palate
- ☐ Clubfoot (talipes equinovarus)
- ☐ Polydactyly
- ☐ Syndactyly
- ☐ Other _____

Growth

- ☐ Short stature
- ☐ Delayed growth
- ☐ Overgrowth
- ☐ Other _____

Perinatal Background

- ☐ Cystic hygroma
- ☐ Non-immune hydrops fetalis (NIHF)
- ☐ Oligohydramnios
- ☐ Premature
- ☐ Polyhydramnios
- ☐ Intrauterine growth restriction (IUGR)
- ☐ Increased nuchal translucency
- ☐ Other _____

Neurodevelopment

- ☐ Intellectual deficiency
  Severity: _____
- ☐ Encephalopathy
- ☐ Developmental delay
- ☐ Language delay
- ☐ Developmental regression
- ☐ Delayed motor development
- ☐ ADHD
- ☐ Autism spectrum disorders autista
- ☐ Others _____

Metabolic

- ☐ Lactic acidosis
- ☐ Organic aciduria
- ☐ High levels of alanine
- ☐ CPK abnormalities
- ☐ Ketosis
- ☐ Low carnitine levels in plasma
- ☐ High levels of pyruvate
- ☐ Other _____

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Clinical features (2)

Ophthalmology

- ☐ Abnormal eye movement
- ☐ Optic atrophy
- ☐ Cataracts
- ☐ Blindness
- ☐ Coloboma
- ☐ Chronic progressive external ophthalmoplegia
- ☐ Ophthalmoplegia
- ☐ Ptosis
- ☐ Retinitis Pigmentosa
- ☐ Nistagmus
- ☐ Strabismus
- ☐ Blurry vision
- ☐ Other _____

Endocrinology

- ☐ Diabetes Mellitus (tipo: ___)
- ☐ Hyperparathyroidism
- ☐ Hypoparathyroidism
- ☐ Hyperthyroidism
- ☐ Hypothyroidism
- ☐ Paraganglioma
- ☐ Pheochromocytoma
- ☐ Other _____

Neurology

- ☐ Ataxia
- ☐ Chorea
- ☐ Seizures/Epilepsy
  (type: _____)
- ☐ Encephalopathy
- ☐ Spasticity
- ☐ Hypotonia
- ☐ Hypertonia
- ☐ Exercise intolerance/fatigue
- ☐ Cerebral palsy
- ☐ Psychiatric disorders
  Specify _____
- ☐ Other _____

Cardiology

- ☐ Arrhythmia/Conduction defect
- ☐ Angioedema
- ☐ Cardiomyopathy
- ☐ CIA
- ☐ VIC
- ☐ Aortic coarctation
- ☐ Left heart hypoplasia
- ☐ Stroke
- ☐ Malformation of the heart and/or great vessels
- ☐ Tetralogy of Fallot
- ☐ Other structural defects of the heart
  Specify _____
- ☐ Other heart abnormalities
- ☐ Specify _____

Gastrointestinal

- ☐ Chronic diarrhoea
- ☐ Hirschsprung disease
- ☐ Constipation
- ☐ Pyloric stenosis
- ☐ Liver failure
- ☐ Tracheoesophageal fistula
- ☐ Gastroschisis
- ☐ Hepatomegaly
- ☐ Omphalocele
- ☐ Chronic intestinal pseudo-obstruction
- ☐ Gastroesophageal reflux
- ☐ Delayed gastric emptying
- ☐ Elevated transaminases
- ☐ Recurrent vomiting
- ☐ Other _____

Nefrología/Genitourinario

- ☐ Kidney agenesis or dysgenesis
- ☐ Cryptorchidism
- ☐ Ambiguous genitalia
- ☐ Hydronephrosis
- ☐ Hypospadias
- ☐ Infertility
- ☐ Kidney malformation
- ☐ Specify _____
- ☐ Renal Tubulopathy
- ☐ Kidney cysts
- ☐ Other _____

Haematology/Immunology

- ☐ Anaemia
- ☐ Coagulation disorders
- ☐ Immunodeficiency
- ☐ Myelofibrosis
- ☐ Neutropenia
- ☐ Pancytopenia
- ☐ Thrombocytopenia
- ☐ Others _____

Dermatology

- ☐ Ampoules
- ☐ Connective tissue abnormalities
- ☐ Hair abnormalities
- ☐ Nail abnormalities
- ☐ Pigmentation abnormalities
- ☐ Ichthyosis
- ☐ Skin tumours
- ☐ Other _____

ENT

- ☐ Hearing loss
  Specify _____
- ☐ External anatomical ear malformations
- ☐ Others _____

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