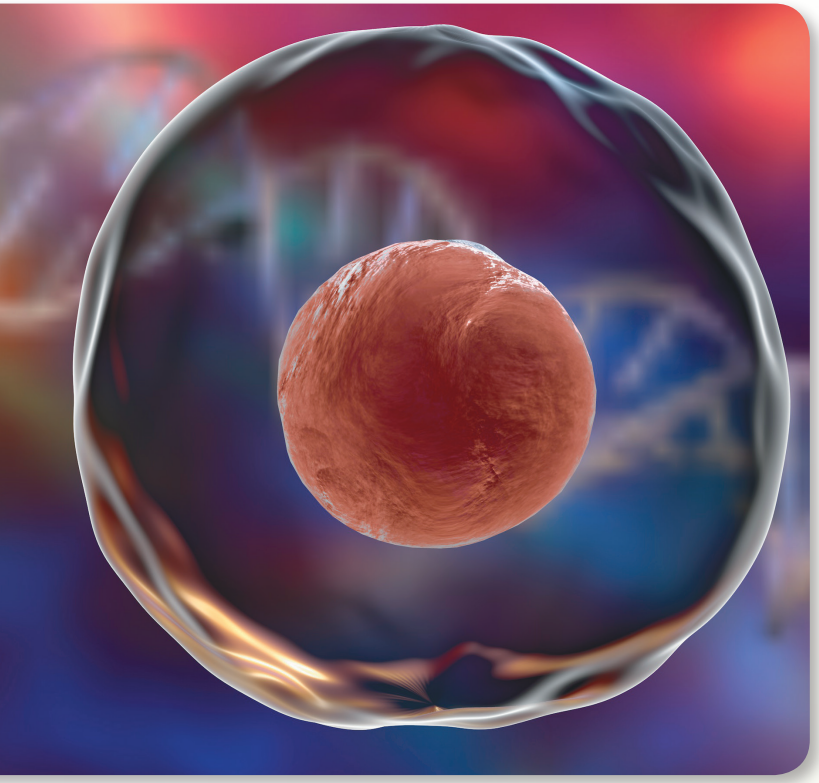
ReproNIM® Focus-LX

For your future

NIMGenetics
New Integrated Medical Genetics

Why ReproNIM® Focus - LX



- Diseases were selected based on their clinical relevance, incidence and scientific societies recommendations.

- Informs on pathogenic and likely pathogenic variants in the genes studied.

- Hand in hand with NIMGenetics, a leading company in genetic diagnosis:
  - Experts in genomics with more than 40,000 studies conducted.
  - Members of the Spanish Association of Prenatal Diagnosis and the Spanish Association of Human Genetics.
  - Clinical reports rated as excellent by opinion leaders in clinical genetics and other medical specialties.

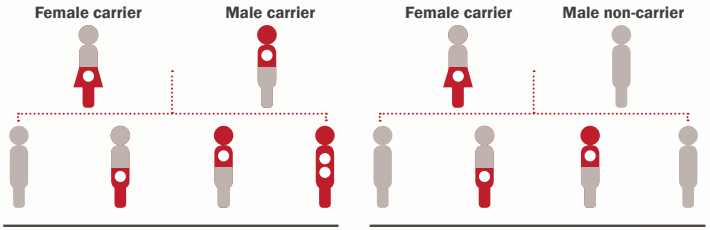
Genetic compatibility test

By making an appropriate selection of the diseases to study and the variants to test, this study minimizes the risk of having an affected child and brings peace of mind to the family.

Parents are the carriers of the genetic information they transmit to their children. The health of children is a fundamental concern of all parents.

By examining for mutations in genes associated with diseases with an autosomal recessive or X-linked inheritance pattern in the parents, it is possible to establish the probability of transmission of genetic diseases.

AUTOSOMAL RECESSIVE INHERITANCE

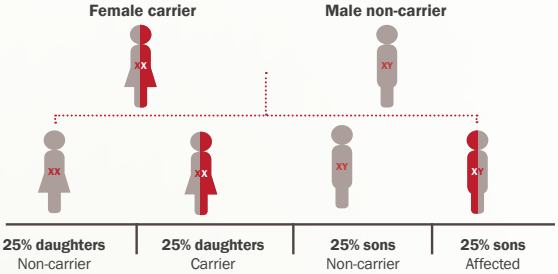


- **50% of children** are carriers
- **25% of children** are non-carriers
- **25% of children** are affected

- **50% of children** are carriers
- **50% of children** are non-carriers

In exceptional circumstances, the appearance of "*de novo*" mutations in the child could determine the development of the disease, irrespective of the carrier status of the parents.

X-LINKED INHERITANCE



Male children who carry the mutation will develop the disease. Daughters who are carriers could present some of the clinical manifestations.

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- **Genetic risk test** that combines different molecular diagnostic techniques for the study of **38 recessive and X-linked diseases.**
- Aimed at knowing the carrier status of mutations in genes in biological progenitors that are associated with genetic diseases.

THIS TEST ANALYZES:

Mutations associated with 38 diseases:

- Clinically relevant
- Early onset
- With the greatest frequency of carriers in the healthy population
- Selected following the recommendations of scientific societies

INDICATED IN THE STUDY OF:

- **Egg and sperm donor**
- **Egg and sperm recipient**
- **Couples who want to be parents**

Genomics
to ensure the future

List of recessive genetic syndromes included in the test:

Autosomal Recessive Disorder	#OMIM	Gene	*OMIM	Prevalence of carriers [a]
Acetyl-CoA dehydrogenase deficiency (medium chain)	201450	*ACADM*	607008	1:62
Canavan disease	271900	*ASPA*	608034	1:71
Cystic fibrosis	219700	*CFTR*	602421	1:28
Smith-Lemli-Opitz syndrome	270400	*DHCR7*	602858	1:68
Pompe disease	232300	*GAA*	606800	1:82
Gaucher disease	230800	*GBA*	606463	1:76
DFNB1 Nonsyndromic hereditary deafness	220290	*GJB2*	212011	1:42
Alpha Thalassemia[3]	604131	*HBA1/HBA2*	141800/141850	1:50
Sickle cell anaemia	603903	*HBB*	141900	1:69
Beta thalassemia	613985	*HBB*	141900	1:69
Tay Sachs	272800	*HEXA*	606869	1:90
Riley-Day syndrome (Familial Dysautonomia)	223900	*IKBKAP*	603722	1:76
Familial Mediterranean fever	249100	*MEFV*	608107	1:64
Phenylalanine hydroxylase deficiency (Phenylketonuria)	261600	*PAH*	612349	1:75
Spinal muscular atrophy[2]	253300	*SMN1*	600354	1:57

(1): Pan-ethnic population data. Lazarín *et al, Genetics in Medicine*, 2013.

(2): Detection of exon 7 deletion by q-PCR.

(3): Detection of the most common deletions/duplications by MLPA.

List of X-linked genetic syndromes included in the test:

By including 23 genes located in the X chromosome, this panel increases the safety compared to the minimum 7-gene panel recommended by the Spanish Fertility Society (SEF) for gamete donors.

X chromosome-linked disease	#OMIM	Gene	*OMIM	Disease prevalence
X-linked adrenoleukodystrophy	300100	*ABCD1*	300371	1/42000
Androgen insensitivity	300068	*AR*	313700	1/20,000;1/50,000
Menkes disease	309400	*ATP7A*	300011	1/99,000
Choroideremia	303100	*CHM*	300390	1/50,000
Alport syndrome, X-linked	301050	*COL4A5*	303630	1/50,000
Chronic granulomatous disease	306400	*CYBB*	300481	1/130,000;1/1,000,000
X-linked hypohidrotic ectodermal dysplasia	305100	*EDA*	300451	1/5,000;1/10,000
X-linked Emery-Dreifuss muscular dystrophy	310300	*EMD*	300384	1/99,000
Haemophilia A	306700	*F8*	300841	1/10,000
Haemophilia B	306900	*F9*	300746	1/10,000
Glucose-6-phosphate dehydrogenase deficiency	300908	*G6PD*	305900	1/200
X-linked Charcot-Marie-Tooth type 1	302800	*GJB1*	304040	1/6,700
Fabry disease	301500	*GLA*	300644	1/50,000;1/117,000
Mucopolysaccharidosis type 2	309900	*IDS*	300823	1/99,000-1/170,000

Severe combined immunodeficiency T-B+	300400	*IL2RG*	308380	1/50,000-1/99,000
X-linked centronuclear myopathy	310400	*MTM1*	300415	1/50,000
Lowe Syndrome	309000	*OCRL*	300535	1/500,000
Ornithine transcarbamylase deficiency	311250	*OTC*	300461	1/70,000
X-linked Leigh syndrome	256000	*PDHA1*	300502	1/40,000
X-linked Charcot-Marie-Tooth type 5	311070	*PRPS1*	311850	<1/1,000,000
X-linked retinoschisis	312700	*RS1*	300839	1-9/99,000
Wiskott-Aldrich syndrome	301000	*WAS*	300392	1-10/1,000,000
Fragile X syndrome[4]	300624	*FMR1*	309550	1:170 (Premutation carriers)[5]

(4): Detection of CGG expansion in 5'UTR region by fragment analysis.

(5): Urbano *et al, Genetic Alterations in Oocyte Donors*, 2014.

Conditions and sample shipment:

- **Sample type:** 3-5 mL of blood in EDTA.
- **Documents to be enclosed with the sample:** Informed consent and request form*.
- **Response time:** 20 business days from the reception of the sample.
- **Reference:** REP1004

(*): If you have any questions, please, call at +34 91 037 83 54 or contact with the sales representative.

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La Suma de Todos
Comunidad de Madrid

NIMGenetics is a Genetics Diagnosis center authorized by the Health and Consumer Department for the Community of Madrid, being duly registered under Nº CS10673

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www.nimgenetics.com

GenQA Genomics Quality Assessment
ISO 9001 BUREAU VERITAS Certification
CDTI Centro para el Desarrollo Tecnológico Industrial
Gobierno de España · Ministerio de Economía y Competitividad
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ENAC Ensayos Nº 1212/LE2336
CEQAS Cytogenetic External Quality Assessment Service
EMQN The European Molecular Genetics Quality Network
ERIC european research initiative on CLL