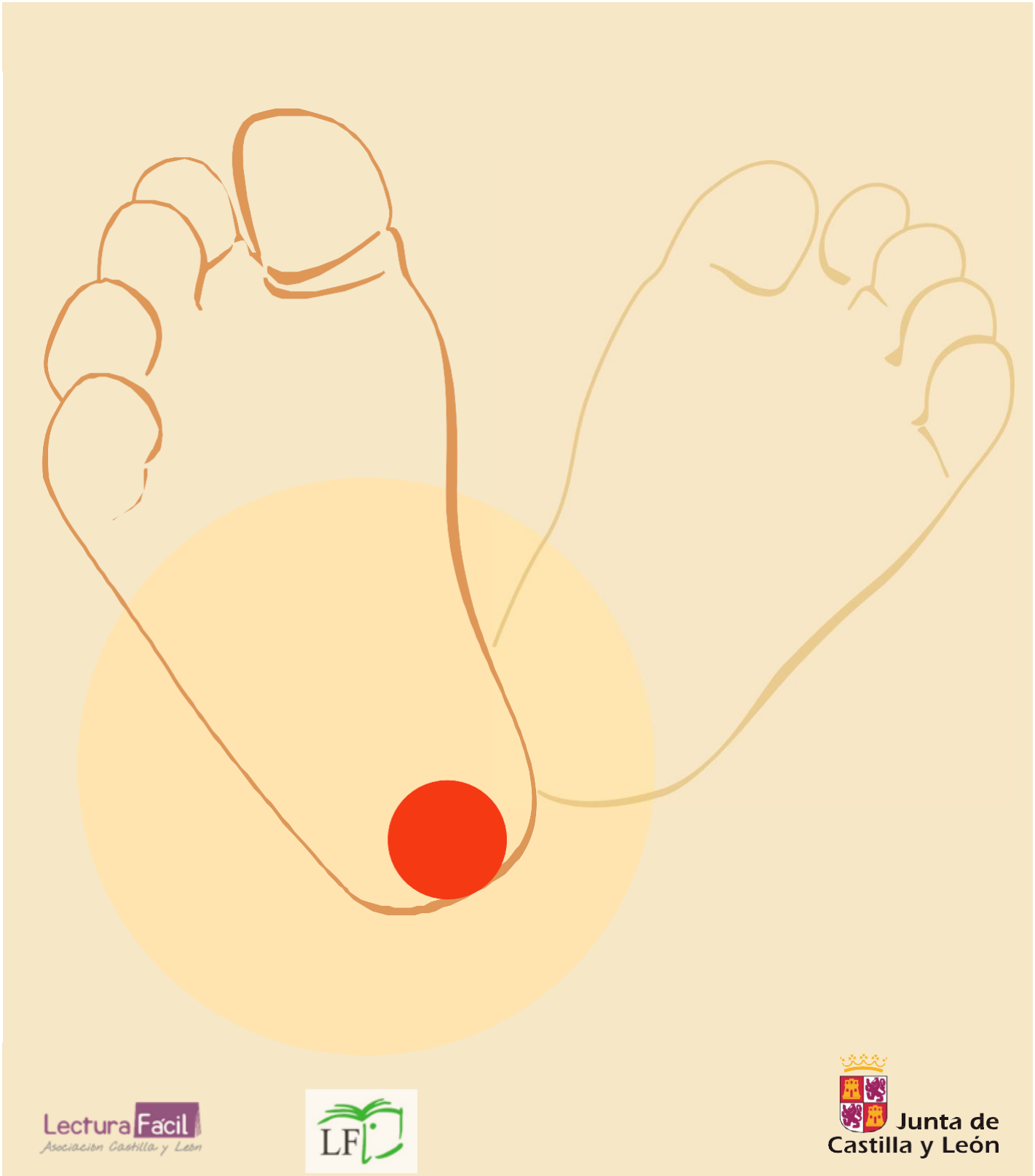
Early detection of congenital diseases



Lectura Fácil
Asociación Castilla y León

LF

Junta de Castilla y León

Early detection of congenital diseases

The Junta de Castilla y León's Regional Health Ministry has a programme for the early detection of congenital diseases in newborns.
This programme allows for the diagnosis and treatment of some diseases that do not cause symptoms in newborns, but which may lead to serious health problems during the first months of their lives.
The programme is voluntary, free-of-charge and available to all new-borns in Castilla y León.

Early detection

Discovering a disease before the symptoms appear.

Congenital diseases

Diseases that are present from the baby's birth and that may or may not be passed on by the parents.

What are congenital diseases?

Congenital diseases are rare diseases because there are only a few cases.
They are a group of diseases with different causes:

- Endocrine: in other words, diseases related to glands and hormones.

Glands are organs that produce substances such as sweat or saliva.

Hormones are chemical substances produced by the body to certain things, such as grow.

- Metabolic: this means diseases related to changes that take place in cells.

- Hematologic: namely, diseases related to components of the blood.

Which diseases does the Programme detect?

The Programme detects 15 different diseases.
Below is a list of their names:

- Congenital hypothyroidism
- Phenylketonuria
- Cystic fibrosis
- Congenital suprarrenal hyperplasia
- Sickle cell disease, Glutaric acidemia type 1
- Medium chain acyl-CoA dehydrogenases deficiency (MCAD),
- Long chain

3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)

- Biotinidase deficiency
- Isovaleric academia
- Maple syrup urine disease
- Homocystinuria
- Tyrosinemia I
- Methylmalonic acidemia
- Propionic Acidemia

Ask your doctor for more information.

What are the advantages of early diagnosis?

Early diagnosis means that the disease can be treated before the child develops symptoms and improves the evolution of the disease.

How, when and where is the test done?

The test consists of extracting a few drops of blood from the baby's heel. The test is done while the baby is still in hospital, on the third day after birth,

when he/she is between 48 and 72 hours old.
The blood is analysed in a laboratory
called the Newborn Screening laboratory located
in the Castilla y León Haemotherapy and Blood Donation Centre.

Newborn screening

a blood test carried out on all newborns for the early detection and treatment of metabolic diseases.

Taking a second sample

Sometimes it is necessary to take a second blood sample.
If this occurs, the laboratory
will inform you by telephone or letter
that you should visit your health centre as soon as possible.

Communication of the results

The Newborn Screening laboratory
will inform you of the results of the test.
If the results are normal,
you will receive a letter 20 days
after the test.

If the letter is late or does not arrive,
you can call 983 418 823
and ask to be put through to extension 89675.
You can call from Monday to Friday
between 11 a.m. and 2 p.m.

If there is anything unusual in the results,
this doesn't mean that your baby has a disease.
More tests will be necessary
before a final diagnosis can be given.

Informed consent and personal data protection

As this is a genetic test,
the mother, father or legal guardian
must give their written consent for the test to be carried out.

The hospital where the test is carried out
will give you all the information you need to know about the following:

- the test
- the consent form you have to sign
- the possibility that you may not be informed
  about some of the Programme's results
- the conservation of the blood sample

and the possibility of you giving your permission
for its use after 5 years,

for research

at the Castilla y León Haemotherapy
and Blood Donation Centre biobank.

Biobank

A place where biological samples are stored for the purpose of diagnosis or research, and which guarantee the quality, order and use of the samples.

The General Directorate of Public Health
is the controller for the processing and use
of your personal data and those of the newborn.

You may exercise your data protection rights
before the General Directorate of Public Health,
or lodge a complaint before the Spanish Data Protection Agency.
If you wish to know more
about how your data are processed, visit
https://www.saludcastillayleon.es/transparencia/es/pdsn0025

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designed for people with reading comprehension difficulties.
It has been validated by persons with intellectual disabilities.
It has been approved by the Castilla y León Easy-to-Read Association.

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