SOURCES: IPOPI, PID UK, EC

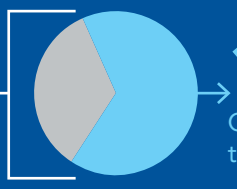
HEALTH FIRST EUROPE

POWER OF KNOWLEDGE: SAVING LIVES OF NEW-BORNS AFFECTED BY SCID

DEFINITION

Primary immunodeficiency diseases (PIDs) are a group of more than 350 rare, chronic disorders in which part of the body's immune system is missing or functions improperly. While not contagious, these diseases are caused by hereditary or genetic defects. Although some disorders are present at birth or in early childhood, the disorders can affect anyone, regardless of age or gender.

19.355
registered patients in the EU (June 2014)



12.775
Children younger than age 15 years

Severe Combined Immune Deficiency (SCID) is the most severe form of primary immunodeficiency (PID) condition. Affected children have very little or no immune system. They are therefore highly susceptible to bacterial, viral, fungal and opportunistic infections. The treatment and prevention of such infections may prolong life but this does not cure SCID.

SCID is also the only form of PIDs that can be cured with treatments. Rapid access to therapy within the first 3.5 months of life has been shown.

>90%
survival rate

BURDEN

Undiagnosed or late-diagnosed Primary Immunodeficiency diseases represent an enormous unmet medical need and a major challenge for public health.

SCID has a major impact upon a child and their family.

100%
mortality of children within the first 2 years of their life

Outlook for siblings

There is strong evidence that in families with multiple cases of SCID the outlook for the firstborn child is significantly worse than that for any subsequent children. This is because the early diagnosis of SCID in second/consequent children allows definite treatment before the onset of a potential severe infection.

THE VALUE OF SCREENING AND EARLY DIAGNOSIS

IT IS ESTIMATED THAT AROUND 60% OF PIDS CAN BE EASILY DIAGNOSED WITH A SIMPLE AND INEXPENSIVE BLOOD TESTS, BUT MANY PIDS REMAIN UNDERDIAGNOSED ON A GLOBAL SCALE.

NEW-BORN SCREENING FOR SCID is crucial to ensure babies can be diagnosed as early as possible and access life-saving treatment rapidly. The chances of curing babies with SCID decrease with time.

€50.000-100.000 cost reduction per case

EARLY DETECTION OF SCID could reduce the cost of treatment by €50.000-100.000 per case. According to a recent a recent Swedish study, the total savings per child was well over €120.000 comparing the mean cost per child of €301.832 for babies who accessed their treatment early and €423.642 for those accessing treatment late (after 6 months of age).

SCID NEW-BORN SCREENING is established in the USA, Israel, New Zealand, and Norway and several EU Member States are currently running national pilot programmes.

POLICY RECOMMENDATIONS

- **INCLUDING AND PERFORMING SYSTEMATIC SCREENING** programmes at birth for SCID;
- **RAISE AWARENESS** at EU and national level by means of educational campaigns and materials to ensure SCID and other PID patients can be diagnosed and treated.
- **FACILITATE ACCESS** to early diagnosis and specialist care.
- **INVESTING IN NEW THERAPIES** for rare diseases and methodologies to assess the impact of diagnoses and therapies on rare disease patients;
- **CONSIDER NEW-BORN SCREENING** as an intrinsic key component of rare diseases care to reduce discrepancies in the European Union between Member States in terms of access to new-born screening.