ESID Registry of Primary Immunodeficiencies

REPORID REGISTO PORTUGUÊS DE IMUNODEFICIÊNCIAS PRIMÁRIAS Nº REPORID _____

Patient (first, middle, last; initials only) escrever todas as iniciais _____ Date of birth (Y/M/D) _____ ☐ Male ☐ Female

Referring physician _____ Tel _____ Fax _____

Address _____

Centro de procedência _____

Antibody deficiencies:

- ☐ Agammaglobulinemia (IgM ____ g/l, IgG ____ g/l, IgA ____ g/l at diagnosis)
  - ☐ X-linked (XLA/Bruton's disease, btk deficiency)
  - ☐ Autosomal recessive
  - ☐ Sporadic
  - ☐ Transient Agammaglobulinemia of infancy
- ☐ Common variable immunodeficiency (IgM ____ g/l, IgG ____ g/l, IgA ____ g/l at diagnosis)
  - ☐ Associated with thymoma
  - ☐ Associated with transcobolamine II deficiency
  - ☐ Other (specify) _____
- ☐ Hyper IgM syndrome (IgM ____ g/l, IgG ____ g/l, IgA ____ g/l at diagnosis)
  - ☐ X-linked
  - ☐ Autosomal recessive
  - ☐ Sporadic
- ☐ Immunoglobulin class deficiency (IgM ____ g/l, IgG ____ g/l, IgA ____ g/l at diagnosis)
  - ☐ Associated with IgG subclass deficiency (specify) _____
- ☐ Immunoglobulin subclass deficiency
  - ☐ IgG subclass (IgG1 ____ g/l, IgG2 ____ g/l, IgG3 ____ g/l, IgG4 ____ g/l at diagnosis)
  - ☐ IgA subclass (IgA1 ____ g/l, IgA2 ____ g/l at diagnosis)
  - ☐ Associated with gene deletion
- ☐ Immunoglobulin light chain deficiency
- ☐ Deficiency of specific antibodies (specify) _____ Português

T cell or combined (T and B cell) deficiencies:

- ☐ Absence of T cells with normal or increased B cells
  - ☐ X-linked severe combined immunodeficiency (IL-2 receptor γ-chain deficiency)
  - ☐ Autosomal recessive
  - ☐ Sporadic
- ☐ Autosomal recessive SCID (absence of T and B cells)
- ☐ Other forms of SCID/CID (specify) _____ Português _____
- ☐ T cell receptor deficiency (specify) _____ Português _____
- ☐ DiGeorge anomaly
- ☐ Ataxia telangiectasia
- ☐ Other chromosomal breakage syndrome (specify) _____ Português _____
- ☐ Deficiency of HLA expression (specify) _____ Português _____
- ☐ Adenosine deaminase (ADA) deficiency
- ☐ Purine nucleoside phosphorylase (PNP) deficiency
- ☐ Wiskott Aldrich syndrome
- ☐ Omenn's syndrome
- ☐ Reticular dysgenesis
- ☐ Short limbed dwarfism
- ☐ Chronic mucocutaneous candidiasis

Continues

Phagocytic disorders:

- ☐ Primary neutropenia (neutrophil count ..... x10^9/l at diagnosis)
  - ☐ Kostmann's disease
  - ☐ Other (specify) ..... Português .....
- ☐ Leukocyte adhesion defect
  - ☐ LAD I (LFA-1 deficiency)
  - ☐ LAD II (Sialyl Lewis X deficiency)
- ☐ Chediak-Higashi syndrome
- ☐ Chronic granulomatous disease
  - ☐ X-linked
  - ☐ Autosomal (specify subcomponent) .....
- ☐ Immunodeficiency with partial albinism
- ☐ Congenital asplenia
- ☐ Other (specify) ..... Português .....

Complement deficiencies:

☐ C1q ☐ C1r ☐ C1s ☐ C2 ☐ C3 ☐ C4 ☐ C5 ☐ C6 ☐ C7 ☐ C8 ☐ C9 ☐ Properdin

☐ Factor B ☐ Factor D ☐ C1-esterase inhibitor deficiency ☐ C3b-inactivator deficiency

Others:

- ☐ X-linked lymphoproliferative syndrome
- ☐ Hyper IgE syndrome (Job's syndrome) (IgE ..... kU/l at diagnosis)
- ☐ Interleukin deficiency (specify) ..... Português .....
- ☐ Interleukin receptor deficiency (specify) .....
- ☐ Other (specify) ..... Português .....

Therapy:

- ☐ Gammaglobulin replacement therapy (please indicate current therapy)
  - ☐ Intravenously
  - ☐ Intramuscularly
  - ☐ Subcutaneously
- ☐ Lymphokine therapy
  - ☐ γ-interferon
  - ☐ IL-2
  - ☐ G-CSF
  - ☐ Other (specify) ..... Português .....
- ☐ Transplantation
  - ☐ Bone marrow transplantation
  - ☐ Other (specify) ..... Português .....
- ☐ Other (specify) ..... Português .....

Family history of immunodeficiency or autoimmunity:

☐ No ☐ Yes (specify) Es pecificar Português

.....

.....

.....

Início dos sintomas (data) ..... Data do diagnóstico .....

Saída : ☐ S ☐ N Data ..... e motivo de abandono .....

Signature ..... Date .....

avor remeter a:
Centro Coordenador do REPORID (Registo Português de Imunodeficiências Primárias)
Serviço de Imunologia, Hospital Geral de Santo António
(instalações do ex-CICAP) Rua D. Manuel II, 4050, Porto

Telefone/Fax: 351 22 6064697 (linha directa)
e-mail: imunologia.hgsa@mail.telepac.pt
imunologia@hgsa.min-saude.pt