א' ניסן תשע"ד
01 אפריל 2014

הנדון: זכאות בעילת מחלה נדירה

1. להלן רשימה מעודכנת אודות מחלות/תסמונות, ששכיחותן לעת עתה, נמוכה מ-1:100,000.

2. על פי התקנות, בנוסף ובמצטבר לתנאי הסטטיסטי, המחלה חייבת להטיל <u>עומס טיפולי כבד ביותר</u>.

3. זכאות בגין מחלה נדירה, מחייבת עמידה בו זמנית <u>בשני התנאים</u> דלעיל.

4. תנאי אחד מן השניים, לא מזכה בגמלה.

5. <u>רשימה זו, מבטלת כל הרשימות הקודמות</u>.

AADC – Aromatic Amino Acid Decarboxylase Deficiency
AIDS
Allagile
Alstrom Syndrome
Asphyxiating Thoracic Dystrophy (see also Jeune's)
Barth Syndrome (3-Methyl Glutaconic Aciduria type II)
Bartter
Bickel Fanconi Syndrome (<u>Completely different from Fanconi anemia</u>)
<u>Blackfan Diamond Anemia</u>
Byler Dis.
C syndrome- Opitz Trigonocephaly
Carbamoyl phosphate synthetase 1 deficiency
Chronic Granulomatosis
Cockayne
Congenital dyserythropoiesis –see also Crigler Najar
Congenital insensitivity to pain with anhydrosis (CIPA)
Crigler Najar -see also congenital dyserythropoiesis
Cyclic Neutropenia
Cystinosis
Donohue Syndrome (Leprechaunism)
Drash
Dyggve Melchior Clausen
Dystonia Ziehen Oppenheim
Ectopia cordis
Ectrodactyly ectodermal dysplasia clefting Syndrome (EEC)
Ehlers Danlos type IV
Eosinophilic Fasciitis
Fibrodysplasia Ossificans Progressiva

Floating Harbor

Freeman Sheldon (see also Whistling face Syndrome)
Glutaric Aciduria type II
Gorham disease
HHH Syndrome
Histiocytosis X
Hunter
Hypercholesterolemia Familial Homozyogtic
Hyper IgE (Job's)
Hyperoxaluria type I
Ichtyosis:(specific types only) Lamellar, Non-bullous Congenital, Ichtyosis erythroderma
Incontinentia Pigmenti
Infantile Polyartheritis Nodosa
Isovaleric Acidemia
Jacobsen Syndrome
Jeune's (Asphyxiating Thoracic Dystrophy)
Job's (Hyper IgE)
Kearns Syre
Kenny-Caffey Syndrome
Kostman Disease
Krabbe Disease
LADD Lacrimo-auricular-dento-digital Syndrome
Langerhans Cell Dis (see also Histiocytosis X)
Larsen Syndrome
Leucocyte Adhesion Deficiency Type 2
Leucodystrophy Van der Knapp
Lymphoma congenital
Morquio Syndrome
Moya-Moya Disease
Mucopolysaccharidosis type II, type IVa, type IVb & type VI
Myhre Syndrome
Nephrogenic Diabetes Insipidus (only X-linked N.D.I)
Netherton Syndrome
Omenn
Pallister-Hall Syndrome
PAN - Periartheritis Nodosa
Papillorenal Syndrome (Pax2 gene mutation)
Pearson marrow pancreas syndrome
Pelizaeus-Merzbacher Disease
Progyria
Pseudo-hypo-aldosteronism
PTLD (post transplantation lymphoproliferative dis)
Pycnodysostosis
Pyruvate Dehydrogenase deficiency
Rogers Syndrome
Schimke Immuno-Osseous Dysplasia
Spondyloepiphyseal Dysplasia
Stevens Johnson
Systemic Scleroderma
Takayasu Vasculitis
Tyrosinemia type I

Tyrosinemia type II
Upshaw Schulman Syndrome
Van der Knapp (see also Leucodystrophy)
Wegener Granulomatosis
Widemann Rautenstrauch Syndrome
Wiskott Aldrich
Xeroderma Pigmentosum
Ziehen Oppenheim Dystonia