
Care
for RARE
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Philippine Society for Orphan Disorders, Inc.

HOPE for POMPE

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Dickoy Magdaraog

Pompe Disease

Description

Pompe disease is a rare neuromuscular genetic disorder that occurs in babies, children, and adults who inherit a defective gene from their parents. There is a defect in a gene that is responsible for making an enzyme called acid alpha-glucosidase (GAA) which is either missing or in short supply.

Patients suffer progressive and debilitating muscular weakness resulting in severe physical disability and dependence on ventilatory support system. The heart and lungs eventually become weak and patients finally succumb to heart and/or pulmonary failure.

Treatment:

Enzyme Replacement Therapy (ERT)

Longevity w/out meds:

infantile form - 12 months
Delayed onset - 2nd-3rd decade of life

Prevalence: *(Documented)*

Worldwide - 1: 40,000
Philippines - 2 cases

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I am RARE

Shaina B. Tayag

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PKU - *secondary to* 6 Pyruroyl tetrahydrobiopterin(6PTPS) deficiency

Description

PKU is a rare inherited metabolic disease that is characterized by levels of the amino acid phenylalanine. If left untreated, excess levels of phenylalanine can cause mental retardation, seizures, movement disorders, and other serious health problems. The earlier these children are diagnosed and treated, the less risk of permanent damage.

TREATMENT:

1) Tetrahydrobioptenn
2) Levo-dopa
3) 5-Hydroxytryptophan
4) Therapy
5) Anticonvulsants

Prevalence: *(Documented)*

Worldwide - 1:10-20,000
Philippines - 1:109,000
(based on newborn screening data for PKU)
6 PTPS deficiency: 2 cases

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MIRACLES come my way

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lana Cadhit

CHARGE Syndrome

Description

CHARGE syndrome is a rare genetic condition that involves multisensory impairment and occurs in 1 out of 12,000 live births. CHARGE is an acronym for a combination of birth "defects", namely:

C – Coloboma or underdevelopment of the eye
H – Heart problems or defects
A – Atresia of the choanae (abnormal structure of the inner nose and breathing anatomy)
R – Retardation of growth
G –Genital abnormalities
E – Ear defects and hearing loss

Treatment:

Coloboma – cataract surgery
Heart problems – PDA surgery
Atreasia Choanae – Surgery
Retardation of growth & development – Physical, occupational, and speech therapies
Genital abnormalities – Maintenance medication and routine urine C/S (culture/sensitivity) or analysis of microorganisms in the urine
Ear problems - Cochlear implant

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I am
Limited
Edition

ALDRIN LUIS ANGCAO

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Urea Cycle Disorders

Description

Urea Cycle Disorders are inherited genetic defects. Normally, the liver supplies six enzymes to metabolize the protein that we eat. In these disorders, the liver fails to produce all of the enzymes necessary to metabolize protein. Instead, one of the enzymes is either partially produced, or not produced at all. The protein then builds up in the bloodstream as ammonia, causing irreversible brain damage, coma, and eventually death. The treatment of urea cycle disorders consists of reducing the amount of protein the child consumes, giving medication to take extra ammonia out of the body, and adding arginine or citrulline (according to the specific UCD being treated) to help the body use protein.

Prevalence: *(Documented)*

Worldwide	Type I	1: 57,000
	Type II	1: 100.000-230,000
Philippines		6 cases

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One day at a TIME

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Rollaine Lindog & Ralph Tristan Valencia

Galactosemia

Description

Galactosemia is a hereditary disease that is caused by the lack of a liver enzyme required to digest galactose.

Treatment:

Galactose restricted diet

Longevity w/out meds:

reach adulthood but become mentally retarded if not diet restricted by 3 weeks, may die at 1-2 weeks from severe gram negative or liver failure bacteria infections

Prevalence: *(Documented)*

Worldwide - 1: 7,500-80,000
Philippines - 1: 68,637 (9 cases)

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Inspite of three I still have HOPE

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Genevieve & Angelo Marco

Maple Syrup Urine Disease (MSUD)

Description

Maple Syrup Urine Disease (MSUD) is a metabolic disease that causes branched chain amino acids (leucine, isoleucine, & valine) to accumulate in the body, creating a toxic effect that can lead to brain swelling, neurological damage, and death. The disease derives its name from the sweet smell of the urine.

Treatment

1.MSUD Formula (3 to 4 days per can)
2.Plasma Amino Acid test (at least once a month)
3.Leucine level (at least once a month)
4.Therapy

Longevity w/out meds:

7 – 10 days

Prevalence: *(Documented)*

Worldwide - 1:185,000-225,000
Philippines - 91

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Our tomorrows are Brighter because you CARE

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Twins Fatima Ligsay & Erika Sedeño

Gaucher Disease

Description

Gaucher Disease is an inherited illness caused by a mutation in the glucocerebrosidase gene leading to the deficiency of glucocerebrosidase. Glucocerebrosidase is an enzyme that breaks down a particular type of fat cell called glucocerebroside.

Deficiency of the glucocerebrosidase enzyme leads to the accumulation of glucocerebrosides in the brain, liver, spleen, skeleton, and other parts of the body leading to dysfunction of these organs.

Treatment:

Enzyme Replacement Therapy (ERT)

Longevity w/out meds:

Type I 6-80 yrs old
Type II 2 yrs old
Type III 2-60 yrs old

Prevalence: *(Documented)*

Worldwide 1:40,000 – 100,000
Philippines 5 cases

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Everyone has a right to quality LIFE

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Jerick Duran

Hunter Syndrome (MPS)

Description

Hunter Syndrome (MPS) also known as mucopolysaccharidosis II (MPS II), is a rare inborn error of metabolism characterized by deficiency of an enzyme known as iduronate sulfatase. This enzyme breaks down specific long sugar molecules in the body called glycosaminoglycans (GAGs) that are found in many parts of the body such as the bone, joints, heart valves, etc.

Deficiency of iduronate sulfatace leads to the accumulation of GAGs in the above organs causing their dysfunction.

Treatment :

1. Enzyme Replacement Therapy (ERT)
2. Supportive management of complications such as: pneumonia, obstructive sleep, apnea, ear infections, etc.
3. Rehabilitation therapy

Longevity w/out meds:

15-20 years old

Prevalence: *(Documented)*

Worldwide - 1: 40,000
Philippines - 21 cases

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Hiling namin sa Maykapal na may mapagpalang kamay na tumulong

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Edwin John Recede

Adrenoleukodystrophy (ALD)

Description

Adrenoleukodystrophy (ALD) is a rare, genetic disorder characterized by the breakdown or loss of the myelin sheath surrounding the nerve cells in the brain and progressive dysfunction of the adrenal gland.

Treatment:

Bone marrow transplantation, Lorenzo's oil, & steroid replacement

Longevity w/out meds:

Death usually occurs within 1 to 10 years after the onset of symptoms.

Prevalence: *(Documented)*

Worldwide - 1: 17,6000
Philippines - 6 cases

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Sabihin sa lahat ng TUMULONG Salamat

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Vanessa Dee

Wilson Disease

Description

Wilson disease is an inherited disorder in which excessive amounts of copper accumulate in the body, particularly in the liver, brain, and eyes. Typically, signs and symptoms of Wilson disease first appear between the ages of 6 and 40, but most often begin during the teenage years.

Liver disease is usually the initial feature of Wilson disease in people between the ages of 6 and 45. Signs and symptoms of liver disease include yellowing of the skin or the whites of the eye (jaundice), fatigue, loss of appetite, and abdominal swelling. Psychiatric or nervous system problems commonly occur in young adults with Wilson disease. Signs and symptoms of these problems can include clumsiness, trembling, difficulty walking, speech problems, deteriorating school work, depression, anxiety, and mood swings. In many individuals with Wilson disease, copper deposits form a green-to-brownish ring, called the Kayser-Fleischer ring, around the cornea (the front surface of the eye). Abnormalities in eye movements, such as the restricted ability to gaze upwards, may also occur.

Treatment:

Anti copper medicine (Cuprimine, or Syprine, or Galzin) plus a 50mg elemental zinc is to be taken for life.

Longevity w/out meds:

few days

Prevalence: *(Documented)*

Worldwide - 1 to 30,000
Philippines - 30 cases

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