Paediatric Refresher Course
2014

The Rare Disease Society of South Africa

NPO 120-991
PBO







DANGER


Statistics

Rare Diseases South Africa

- If all of the people with rare diseases lived in one country, it would be the world's 3rd most populous country
- Approximately 50% of the people affected by rare diseases are children
- 30% of children with rare disease will not live to see their 5th birthday
- Rare diseases are responsible for 35% of deaths in the first year of life

Statistics

- The prevalence distribution of rare diseases is skewed – 80% of all rare disease patients are affected by approximately 350 rare diseases
- 95% of rare diseases have not one single FDA approved drug treatment
- During the first 25 years of the Orphan Drug Act (passed in 1983), only 326 new drugs were approved by the FDA and brought to market for all rare disease patients combined.
- Approximately 50% of rare diseases do not have a disease specific foundation supporting or researching their rare disease.

RARE DISEASES IN NUMBERS

Rare Diseases South Africa

Preliminary report from an on going bibliographic study initiated by Eurordis in partnership with Orphanet:

Method
Selection of rare disease
(for the purposes of the current report)
- The most common rare diseases according to books and websites
- The most frequently requested pages on the Orphanet website

RARE DISEASES IN NUMBERS

Rare Diseases South Africa

Findings:

- 359 “More common” rare diseases

- Mode of inheritance of 359 rare diseases is:
  - 26.5% autosomal dominant inheritance
  - 28.1% autosomal recessive inheritance
  - 7% X-linked inheritance
  - 10% several modes of inheritance
  - 13.4% multigenic/multifactorial diseases
  - 8.1% sporadic diseases
  - 5.8% unknown aetiology

RARE DISEASES IN NUMBERS

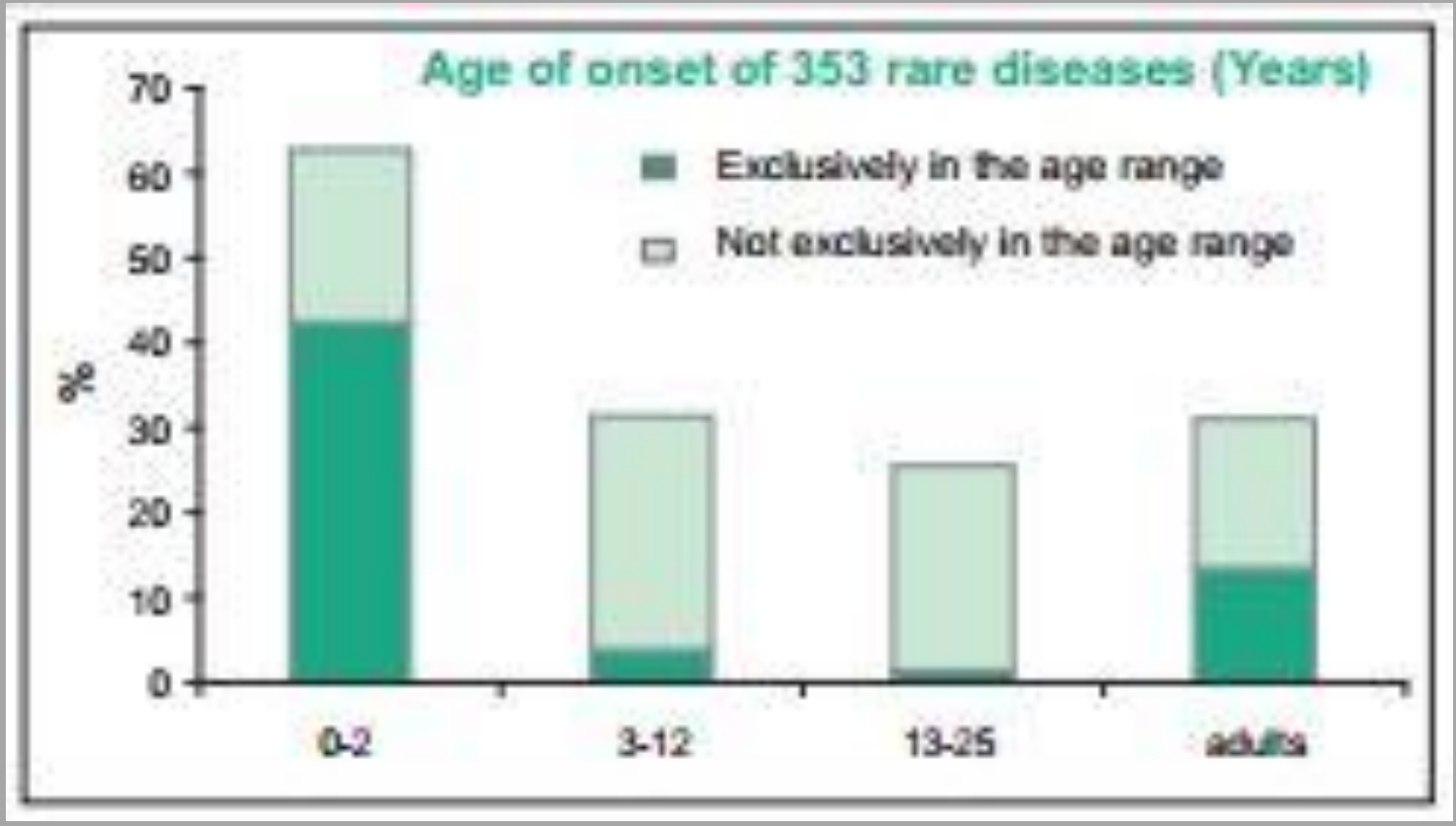
Rare Diseases South Africa

Life Expectancy of these cases:

- - 37.5% normal lifespan
- - 25.7% potentially lethal at birth or before 5 years of age
- - 36.8% reduced lifespan, depending on the severity,
- penetrance or type (child, juvenile or adult types for example) of the disease

RARE DISEASES IN NUMBERS

Rare Diseases South Africa



Disease name	Estimated prevalence (/100 000)
Brugada syndrome	50
Protoporphyria, erythropoietic	50
Guillain-Barre syndrome	47,5
Melanoma, familial	46,8
Autism, genetic types	45
Tetralogy of Fallot	45
Scleroderma	42
Great vessels transposition	32,5
Focal dystonia	30
Marfan syndrome	30
Non-Hodgkin malignant lymphoma	30
Retinitis pigmentosa	27,5
Gelineau disease	26
Myeloma, multiple	26
Alpha-1 antitrypsin deficiency	25
Diaphragmatic hernia, congenital	25
Juvenile arthritis, idiopathic	25
Neurofibromatosis type 1	25
Oesophageal atresia	25
Polycythemia vera	25
Charcot-Marie-Tooth disease	24
Polycystic kidney disease, recessive type	23
VATER association	23
Coffin-Lowry syndrome	22,5
Rendu-Osler-Weber disease	21,25
Dermatitis herpetiformis	20,2
Atresia of small intestin	20
Duodenal atresia	20
Ehlers-Danlos syndrome, classic type	20
Hirschsprung disease	20
Microdeletion 22q11	20
Spherocytosis hereditary	20
Tumor syndrome	20
Cardiomyopathy, familial dilated	17,5
Breast cancer, familial	17
MELAS syndrome	16
Leucinosis	15,6
Acyl-CoA dehydrogenase, medium chain, deficiency of	15
Lennox-Gastaut syndrome	15
Fragile X syndrome	14,25
Primary biliary cirrhosis	13,5
Stickler syndrome	13,5
Williams syndrome	13,3
Willebrand disease	12,5
Gastroschisis	12
Microphthalmia	12
Omphalocele	12
Sarcoidosis	12
MURCS association	11,25
Stargardt disease	11,25
Glioblastoma	11
Multiple endocrine neoplasia type 1	11
Prader-Willi syndrome	10,7
Alopecia totalis	10,5
Nephroblastoma	10,1
Cystic fibrosis	10
Duane syndrome	10
Neuroblastoma	10
Hodgkin disease	9,4

Disease name	Estimated prevalence (/100 000)
Dermatomyositis	9,25
Polymyositis	9,25
Tuberous sclerosis	8,8
Congenital adrenal hyperplasia	8,5
Rett syndrome	8,2
Angelman syndrome	8
Cataract, total congenital	7,9
Hyperlipidemia type 3	7,8
Hemophilia	7,7
Trisomy 18	7,7
Behcet disease	7,5
Immunodeficiency, common variable	7,5
Microscopic polyangiitis	7,5
Idiopathic torsion dystonia	7,25
Oculocutaneous albinism	7,15
Facioscapulohumeral muscular dystrophy	7
Holoprosencephaly	7
Sclerosing cholangitis	7
Sotos syndrome	7
Galactosemia	6,8
Optic atrophy, Leber type	6,5
Osteogenesis imperfecta	6,5
Smith-Lemli-Opitz syndrome	6,5
Amyotrophic lateral sclerosis	6
Treacher-Collins syndrome	6
Tay-Sachs disease	5,75
Christ-Siemens-Touraine syndrome	5,5
Pheochromocytoma	5,5
Retinoblastoma	5,4
Rubinstein-Taybi syndrome	5,4
Alzheimer disease, familial	5,3
Zollinger-Ellison syndrome	5,3
Cornelia de Lange syndrome	5,25
Familial adenomatous polyposis	5,25
Huntington disease	5,25
Acromegaly	5
Fructose intolerance	5
Primary ciliary dyskinesia	5
Supranuclear palsy, progressive	5
Porphyria, acute intermittent	5
Sickle cell anemia	4,8
Deletion 5p	4,6
Myasthenia gravis	4,55
Achondroplasia	4,5
Steinert myotonic dystrophy	4,5
Ceroid lipofuscinosis, neuronal	4
Phenylketonuria	4
Smith-Magenis syndrome	4
Wilson disease	4
Muscular dystrophy limb girdle type 2A, Erb type	3,8
CDG syndroms	3,75
Niemann-Pick A disease	3,75
Propionic acidemia	3,75
Waardenburg syndrome type 1, type2 and type 3	3,75
Beckwith-Wiedemann syndrome	3,65
Adrenoleukodystrophy, X-linked	3,5
Goldenhar syndrome	3,5
Usher syndrome	3,5
Muscular dystrophy, Duchenne and Becker type	3,4
Multiple endocrine neoplasia, type 2	3,3

Disease name	Estimated prevalence (/100 000)
Systemic mastocytosis	3,3
Von Hippel-Lindau disease	3,25
Polyarteritis nodosa	3,07
Friedreich ataxia	3
Poland anomaly	3
Proximal spinal muscular atrophy	3
Saethre-Chotzen syndrome	3
Wegener granulomatosis	3
Kennedy disease	2,8
Cystinosis	2,75
Amaurosis congenita of Leber	2,5
BOR syndrome	2,5
Bullous pemphigoid	2,5
Kartagener syndrome	2,5
Niemann-Pick B disease	2,5
Pseudoxanthoma elasticum	2,5
Leigh disease	2,25
Peutz-Jeghers syndrome	2,2
Autosomal dominant spinocerebellar ataxia	2,15
Albinism ocular	2
Alport syndrome	2
Crouzon disease	2
Deletion 4p	2
Klippel feil syndrome	2
Langerhans cell histiocytosis	2
Nail-patella syndrome	2
Persistent hyperinsulinemic hypoglycemia of infancy	2
Aniridia, sporadic	1,75
Fabry disease	1,75
Variegate porphyria	1,7
Budd-Chiari syndrome	1,5
Darier disease	1,5
X-linked severe combined immunodeficiency, T- B+	1,5
Bile ducts paucity, syndromic form	1,4
Cat-eye syndrome	1,35
Apert syndrome	1,25
Spastic paraplegia, familial	1,25
Adult Onset Still's disease	1,23
Pierre Robin syndrome	1,2
Glycogen storage disease type 2	1,1
Mucopolysaccharidosis type 3	1,1
Zellweger syndrome	1,1
Nephronophtisis	1,05
3-hydroxyacyl-CoA dehydrogenase, long chain, deficiency of	1
Albers-Schonberg disease	1
Angioneurotic edema	1
Ataxia telangiectasia	1
Chondrodysplasia punctata, rhizomelic type	1
Coloboma, ocular	1
Emery-Dreifuss muscular dystrophy, X-linked	1
Fanconi anemia	1
Gaucher disease	1
Gorlin syndrome	1
Holt-Oram syndrome	1
Hypokalemic periodic paralysis	1
Isovaleric acidemia	1
Mucopolysaccharidosis type 1	1
Nemaline myopathy	1
Neuroendocrine tumor	1
Thomsen and Becker disease	1

Disease name	Estimated prevalence (/100 000)
Churg-Strauss syndrome	0,9
Ellis Van Creveld syndrome	0,9
Joubert-Bolthauser syndrome	0,85
Bardet-Biedl syndrome	0,8
Ebstein anomaly	0,75
Hyperkalemic periodic paralysis	0,75
Krabbe disease	0,75
Mucolipidosis type 2	0,75
Albright hereditary osteodystrophy	0,72
Menkes syndrome	0,7
Niemann-Pick C disease	0,7
Glycogen storage disease type 4	0,6
Alpha-sarcoglycanopathy	0,57
Beta-sarcoglycanopathy	0,57
Delta-sarcoglycanopathy	0,57
Gamma-sarcoglycanopathy	0,57
Tetrasomy 18p	0,55
Neurofibromatosis type 2	0,5
Xeroderma pigmentosum	0,5
Agammaglobulinemia X-linked	0,45
Cowden syndrome	0,45
Werner syndrome	0,45
Glutaryl-CoA dehydrogenase deficiency	0,4
Homocystinuria due to cystathionine beta-synthase deficiency	0,4
Mucopolysaccharidosis type 4	0,4
Leach-Nyhan syndrome	0,38
Pfeiffer syndrome	0,38
Severe combined immunodeficiency T- B-	0,35
Anemia congenital hypoplastic, Blackfan-Diamond type	0,32
Alkaptonuria	0,3
Lissencephaly, type 1, due to LIS 1 anomalies	0,3
Lipodystrophy, Berardinelli type	0,25
Progeria	0,25
Granulomatous disease, chronic	0,2
Jeune syndrome	0,2
Nanism due to growth hormone resistance	0,2
Neurodegeneration with brain iron accumulation (NBIA)	0,2
Creutzfeldt-Jakob disease	0,19
Lowe syndrome	0,19
Mucopolysaccharidosis type 6	0,18
CHARGE association	0,14
Metachromatic leukodystrophy	0,13
Bartter syndrome	0,12
Muscular dystrophy fukuyama type	
Walker-warburg syndrome	
Muscle eye brain disease	0,12
Ewing sarcoma	0,1
Hypercholesterolemia, familial (homozygous form)	0,1
Fibrodysplasia ossificans progressiva	0,08
Dopa-responsive dystonia	0,05
Tyrosinemia type 1	0,05
Factor XIII deficiency, congenital	0,04
Perinatal hypophosphatasia	0,03

The biggest challenges?

Rare Diseases South Africa

- Getting an accurate diagnosis
- Assuring patient access to appropriate treatment
- Strengthening ones ability to self-advocate.
- Emotional support

Accurate Diagnosis

Rare Diseases South Africa

Many genetic tests not available locally.

So, we have partnered up with industry leaders DHL who move all our products to testing facilities around the world at no cost.

DHL

DHL

Rare Diseases South Africa

Access to treatment/therapies

Rare Diseases South Africa

Many conditions only have supportive care.

We work closely with the dieticians/therapists and do specific drives to ensure our patients get adequate feed supplementation and therapy.

Access to treatment/therapies

Rare Diseases South Africa

Often access to equipment such as gait walkers/ wheelchairs is not an option, So we have started a “swop Shop” where families can locate these items at a better price, and often parents have wanted to “pay it forward” and give the items away.

Access to treatment/therapies

Rare Diseases South Africa

Financial Assistance For:

Medical aid subsidies

Travel Grants

Specialist appointment fees

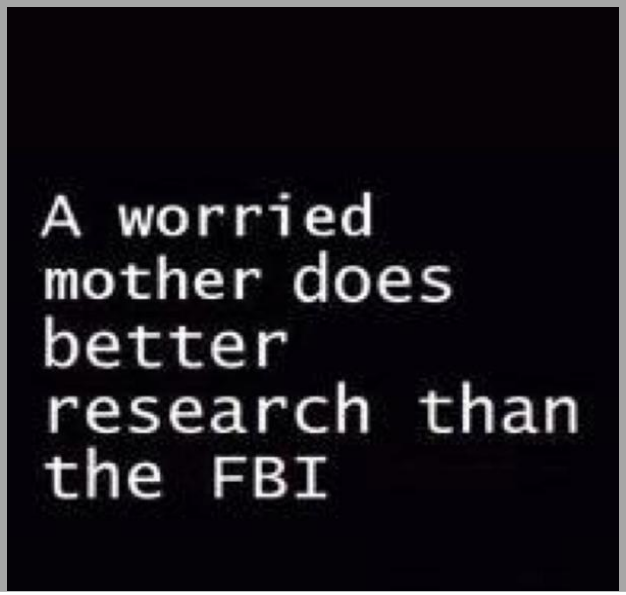
Diagnostics

Self-advocating

Rare Diseases South Africa

"Knowledge is power"

Aim to get parents well educated in terms of the related condition, so that day-to-day living is manageable.



Emotional support…

Rare Diseases South Africa

''Inherited Day"

Genetic counselling/ family counselling.

Ambassador Programme

Our work with healthcare providers…

Rare Diseases South Africa

Includes logistical support in terms of testing, appointments, scripts etc.

Assistance with medical aid authorization

Council for medical scheme follow ups

Conveying of important information to patients

Aim is to start a referral network between specialists and healthcare providers which will assist with sharing of information and experience.

Educating the public…

Rare Diseases South Africa

Implementing Various awareness days

Sharing of important industry news via social media

Telling patient stories to raise awareness for specific conditions

Get involved?

Rare Diseases South Africa

- Sign up to be on our professional network at www.rarediseases.co.za.
- Help us with awareness activities
- Refer patients to our team

Contact Us:

Rare Diseases South Africa

Chairperson
Name: Kelly du Plessis
Mobile no: 072 623 6763
Email: info@rarediseases.co.za

Vice Chair/Fundraising Coordinator
Name: Shevaun McCreedy
Mobile no: 082 415 3893
Email: fundraising@rarediseases.co.za

Administrator/Events Coordinator
Name: Nikki Melless
Mobile no: 083 705 9122
Email: admin@rarediseases.co.za

Follow Us:

Rare Diseases South Africa

Website: www.rarediseases.co.za

Facebook: Rare-Diseases-SA

Twitter: @RarediseasesSA