CDG AWARENESS AND DISSEMINATION KIT :

- Kit materials include:
  - ❑A powerpoint with an introduction about Rare Diseases and Living with a Rare Disease (CDG in our case)
  - ❑Fundraising and Conference presentation course

For CDG AWARENESS AND DISSEMINATION KIT materials, please contact: cdgawareness@gmail.com

CONGENITAL DISORDERS OF GLYCOSYLATION (CDG) AWARENESS AND DISSEMINATION PROJECT

QUESTIONS ABOUT PRESENTATION SUPPORT CONCERNING:

- **INTRODUCTION ABOUT RARE DISEASES (RD)**
- **THE PATIENT'S VOICE**
- **CONFERENCE PRESENTATIONS COURSE**

CONTACT: Vanessa Ferreira (sindromecdg@gmail.com)

- **HAVING A RARE DISEASE: LIVING WITH CDG**

CONTACT: Bas Holten (basesdownunder2003@hotmail.com)

TUTORIAL FOR FUNDRAISING:

- **SIMPLE STEPS FOR FUNDRAISING SUCCESS**

CONTACT: Andrea Berarducci (maui911@yahoo.com)

CONGENITAL DISORDERS OF GLYCOSYLATION (CDG) AWARENESS AND DISSEMINATION PROJECT

In each of the following slides, we suggest some sources of information and ideas about what you may want to discuss and highlight in your presentation…

Be aware that some information should be adapted to the country where you live. Thanks!

Please, all information should be used with respect and integrity. Thanks.

SKYPE FOR HELP IN ORAL PRESENTATION PREPARATION:
Skype name: cdgawareness

PLEASE IF YOU USE THIS AWARENESS MATERIAL WE KINDLY REQUEST YOU TO MENTION AS SOURCE:

CDG AWARENESS DONE BY CDG PATIENT'S VOICE

CONGENITAL DISORDERS OF GLYCOSYLATION (CDG)

Name, CDG patient representative
Country

OUTLINE

- **INTRODUCTION ABOUT RARE DISEASES (RD)**
- **THE CDG PATIENT'S VOICE**
- **LIVING WITH CDG**
- **SIMPLE STEPS FOR FUNDRAISING SUCCESS**

WHAT IS A RARE DISEASE?

A rare disease in Europe is a disease affecting less than 1 in 2,000 citizens

In the United States, a rare disease is any disease or condition that affects 1 in 1,500 people

- 29 million people affected in the EU
  - 3 million people Spain
  - 3 millions people in France (1 in 20)
  - 600 000-800 000 people in Portugal
  - 3.5 million people in the UK
  - 1 million people in the Netherlands

- 25 million people USA

6,000 and 8,000 distinct rare diseases!

49 XXXXY 5p, Síndrome Acidemia Metilmalónica Homocistinuria, Tipo cbl C Acidemia Propiónica Acondroplasia Acondroplasia - Inmunodeficiencia Combinada Grave Adrenoleucodistrofia Agammaglobulinemia Ligada al Cromosoma X Aicardi Goutières, Síndrome de Alagille, Síndrome de Albinismo Alexander, Enfermedad de Alfa 1 Antitripsina, Déficit de Alpers, Enfermedad de Alport, Síndrome de Amaurosis Retiniana Congénita de Leber Amiloidosis Primaria Familiar Andrade, Enfermedad Anemia de Fanconi Angelman, Síndrome de Angioedema Hereditario Aniridia Apert, Síndrome de Arnold Chiari, Síndrome de Arteritis de Células Gigantes Artritis Crónica Juvenil Artritis Idiopática Juvenil Artritis Psoriásica Artrogriposis Múltiple Congénita Artrogriposis, Síndrome de Aspartilglucosaminuria Ataxia de Friedreich Ataxia de Marie Ataxia Espinocerebelosa del Tipo 1 (SCA1) Ataxia Espinocerebelosa SK3 Ataxia Hereditarias Ataxia Olivopontocerebelosa Ataxia Telangiectasia Atresia Aórtica Atresia Pulmonar con Comunicación Interventricular Atresia Pulmonar Septo Ventricular Intacto Atresia Tricúspide Atrofia Muscular Espinal Infantil Atrofia Muscular Espinal Proximal de Tipo 2 Atrofias Espinales Baller Gerold, Síndrome de Batten Spielmeyer Vogt, Enfermedad de Beckwith Wiedemann, Síndrome de Behçet, Enfermedad de Berardinelli Seip, Síndrome de Blefaroespasmo B-Oxidación Mitocondrial Braquicefalia Aislada Budd Chiari, Síndrome de Buerger, Enfermedad de C de Opitz, Síndrome Calambre del Escribano Calcinosis Canavan, Enfermedad de Cardiopatías Congénitas Carnitina, Síndromes por Déficit de Castleman, Enfermedad de Ceroido Lipofuscinosis Juvenil Ceroido Lipofuscinosis Neuronal Charcot Marie Tooth, Enfermedad de Chediak Higashi, Enfermedad de Churg Strauss, Síndrome de Cistinosis Cistinuria Cistitis Intersticial Citocromo C Oxidasa, Déficit de Citomegalovirus, Síndrome del Citrulinemia Coartación Aórtica Coffin Lowry, Síndrome de Coffin Siris, Síndrome de Colangitis Crónica Destructiva no Supurativa (CBP) Colangitis Primaria Esclerosante Coloboma del Iris Complejo Malformativo de Arnold Chiari Coproporfiria Hereditaria Corea de Sydenham Cornelia de Lange, Síndrome de Coroidemia Craneosinostosis Primaria Crecimiento, Problemas de Crigler Najjar, Síndrome de Cromosoma X Frágil, Síndrome de Crouzon, Enfermedad de Dandy Walker, Síndrome de Danon, Enfermedad de Defectos del Tabique Interauricular Defectos en la Biosíntesis de Testosterona Deficiencia Múltiple de Sulfatasas Déficit Congénito de HFE Déficit de 5a-reductora Deformidad de Sprengel Degeneración Macular Denys Drash, Síndrome de Depleccóon del ADN Mitocóndrial, Síndrome Dilatación Aórtica Disferlina, Ausencia de Disfonía Espasmódica Disgenesia Gonodal XY Dismetrías Óseas Displasia Ectodérmica Displasia Ectodérmica Tipo Berlin Displásia Espondofisária Displasia Ósea Displasia Ventricular Derecha Arritmógena (DVDA) Distonia Cervical Distonía Focal Distonía Multifocal Paroxística Distonía Neurodegenerativa Distonía Primaria Distonía que Responde a la Levodopa Distrofia Miotónica Distrofia Muscular Congénita Tipo Fukuyama Distrofia Muscular de Cinturas Distrofia Muscular de Duchenne y de Becker Distrofia Muscular de Emery Dreyfuss Distrofia Muscular de Landouzy Dejerine Distrofia Muscular Infantil Distrofias Musculares Doble Salida de Ventrículo Derecho Dolor Pélvico Crónico Duchenne Erb, Síndrome de Dumping, Síndrome de Ehlers Danlos, Síndrome de Ellis Van Creveld, Síndrome de Enanismo de Laron Epidermolisis Bullosa Epilepsia Mioclónica Progresiva Escafocefalia Esclerodermia Esclerosis Lateral Amiotrófica Esclerosis Tuberosa Espina Bífida Estenosis Pulmonar Valvular Estenosis Subaórtica Fija Exostosis Múltiple Extrofia Cloacal Extrofia Vesical Fabry, Enfermedad de Factor IX, Déficit de Fascitis Eosinofílica Fatiga Crónica, Síndrome de Felty, Síndrome de Fenilcetonúria o PKU Fibrodisplasia Osificante Progresiva Fibrosis Quística Fiebre Reumática Fucosidosis Galactosemia, Enfermedad de Gangliosidosis GM1 Gaucher Schlagenhaufer, Enfermedad de Gilles de la Tourette, Síndrome de Glicosilación, Defectos Congénitos de la Glucogenosis Gorlin, Síndrome de Granulomatosa Cronica, Enfermedad Granulomatosis de Wegener Hemiplejia Alternante Infantil Hemocromatosis Familiar Hemocromatosis Neonatal Hemofilia A Hemofilia C Hemoglobinopatía C - Forma Homocigota Hemoglobinopatía C Harlem Hemoglobinopatía D Hemoglobinopatía E Hemoglobinopatía Inestable Hemoglobinopatía J Hemoglobinopatía M con Hemólosis Hemoglobinopatía S Hemoglobinuria Paroxística Nocturna Hepatitis Autoinmune, Formas Mixtas de Hermansky Pudlak, Síndrome de Hidrocefalia Hiper-IgM, Síndrome de Hiperlaxitud, Síndrome de Hiperlisinemia Hiperostosis Frontal Interna Hipertensión Pulmonar Hipertensión Pulmonar Primaria o Idiopática Hipertensión Pulmonar Secundaria Hipocondroplasia Hipomagnesemia por Malabsorción Selectiva de Magnesio Hipopituitarismo Hipoplasia de las Celulas de Leydig Hipotonía Muscular Congénita Histiocitosis Tipo II Histiocitosis X Hormona del Crecimiento, Déficit de Hunter, Síndrome de Huntington, Enfermedad de Huntington, Enfermedad de (Variante de Wesphal) Hurle Scheie, Síndrome de Hurler, Enfermedad de I - Cell Ictiosis Vulgar Inmunodeficiencia Variable Común Insensibilidad a los Andrógenos, Síndrome de Insuficiencia Suprarrenal Primaria, Crónica, Adquirida Intolerancia Hereditaria a la Fructosa Isaacs, Síndrome de Jacobsen, Síndrome de Job, Síndrome de Joseph, Enfermedad de Joubert, Síndrome de Kawasaki, Síndrome de Kearns Sayre, Síndrome de Kennedy, Enfermedad de Kleine Levin, Síndrome de Klinefelter, Síndrome de Klippel Feil, Síndrome de Krabbe, Enfermedad de Leigh, Síndrome de Lesch Nyhan, Síndrome de Leucemia Linfocítica Crónica Leucodistrofia Leucodistrofia Metacromática Linfangioleiomiomatosis Lowe, Enfermedad de Lupus Discoide Eritematoso Lupus Eritematoso Sistémico Malformaciones Craneocervicales Manosidosis Marfan, Síndrome de Maroteaux Lamy, Síndrome de Marshall Smith, Síndrome de Mastocitosis Maullido del Gato, Síndrome del McArdle, Enfermedad de McCune Albright, Síndrome de McLeod, Síndrome de Melas, Síndrome de MERRF, Síndrome Miastenia Gravis Miller Dieker, Síndrome de Miocardiopatía Dilatada Idiopática Mitocondrial Mioneurogastrointestinal, Síndrome (MNGIE) Miopatía de Multicore Miopatía Mitocondrial Miopatía Nemalínica Miopatías Congénitas Miopatías Metabólicas Mitoconfrial, Enfermedad Mixta del Tejido Conectivo, Enfermedad de Moebius, Síndrome de Morquio, Enfermedad de Mucolipidosis Tipo 2 Musculares, Enfermedades Narcolepsia NARP, Síndrome de Natowicz, Síndrome de Nefronoptisis Neuramidasa, Deficiencia de Neurinoma del Acústico Neurofibromatosis Central Neurofibromatosis de Von Recklinghausen Neuromusculares, Enfermedades Neuropatía Motora Multifocal Nevus Nevus Gigante Congénito Niemann Pick, Enfermedad de Nonne, Síndrome de Noonan, Síndrome de Núcleo Central, Enfermedad de Nutrición Parenteral Oculo Cerebro Renal, Síndrome de Oftalmoplegia Externa Progresiva Osteogénesis Imperfecta Osteonecrosis Panhipopituitarismo Paraparesia Espástica Familiar Paraparesia Espástica Tropical Parry-Romberg, Síndrome de Pearson, Síndrome Pelizaeus Merzbacher, Enfermedad de Pénfigo Benigno Crónico Familiar de Hailey-Hailey Pénfigo Foliáceo Pénfigo Vulgar Penfigoide Ampollar Pénfigoide Bulloso Penfigoide Cicatricial Penfigoide Gestacional Persona Rígida, Sindrome de la Peutz Jeghers, Síndrome de Pfeiffer, Síndrome Picnodisóstosis Pierre Robin, Síndrome de Plagiocefalia Poems, Síndrome de Poland, Síndrome de Poliangeitis Microscópica Polineuropatía Amiloide Familiar (Tipo 1) Polineuropatía sensitivo-motora desmielinizante crónica Polineuropatías Poliquistosis Renal Autosómica Dominante (PQRAD) Poliquistosis Renal Autosómica Recesiva Pompe, Enfermedad de Porfiria Aguda de Doss Porfiria Aguda Intermitente Porfiria Cutánea Tarda Porfiria Eritropoyética Congénita Porfiria Hepática Variegata Post Polio, Síndrome de Prader Willi, Síndrome de Primarios Inmunitarios, Déficit de Prostatitis Crónica Proteus, Síndrome de Protoporfiria Eritropoyética Pseudo Hurler Polidistrofia Pseudoxantoma Elástico Púrpura de Schonlein Henoch Químico Múltiple, Síndrome Quiste Aracnoideo Quiste de Tarlov Rapp Hodgkin, Síndrome de Raynaud, Enfermedad Reiter, Síndrome de Retinosis Pigmentaria Retinosquisis Rubinstein Taiby, Enfermedad de Russell Silver, Síndrome de Sandhoff, Enfermedad de SanFilippo, Síndrome Santavuori, Enfermedad de Schwartz-Jampel, Síndrome de Sialidosis Síndrome de Rett Siringomielia Situs Inversus Cardiopatía Sjögren Primario, Síndrome de Sly, Síndrome de Smith Magenis, Síndrome de Sotos, Síndrome de Stargardt, Enfermedad de Stickler, Síndrome de Still del Adulto, Enfermedad de Takayasu, Enfermedad de Talasemia Mayor Talasemia Minor Tay Sachs, Enfermedad de Telangiectasia Hemorrágica Hereditaria Tetralogía de Fallot Tirosinemia Tipo I Transposición de Grandes Vasos Trisomia Cariotipo 47 XXX Turner, Síndrome de Usher, Síndrome de VACTERL Vasculitis Necrosante Generalizada Vasculitis Predominante Cutánea Ventrículo Único Von Gierke, Enfermedad de Von Hippel Lindau, Síndrome de Von Willebrand, Enfermedad de WAGR, Síndrome de Walker Warburg, Síndrome de West, Síndrome de Williams, Síndrome de Wilson, Enfermedad de Wiskott Aldrich, Síndrome de Wolff Parkinson White, Síndrome de Wolfram, Síndrome de X-Frágil, Síndrome Zellweger, Síndrome de

number of scientific publications about rare diseases continues to increase

CHARACTERISTICS OF RARE DISEASES

- ❑ Chronic
- ❑ Progressive
- ❑ Degenerative
- ❑ Life-threatening
- ❑ Disabling
- ❑ Lack or loss of autonomy
- ❑ Difficult to manage

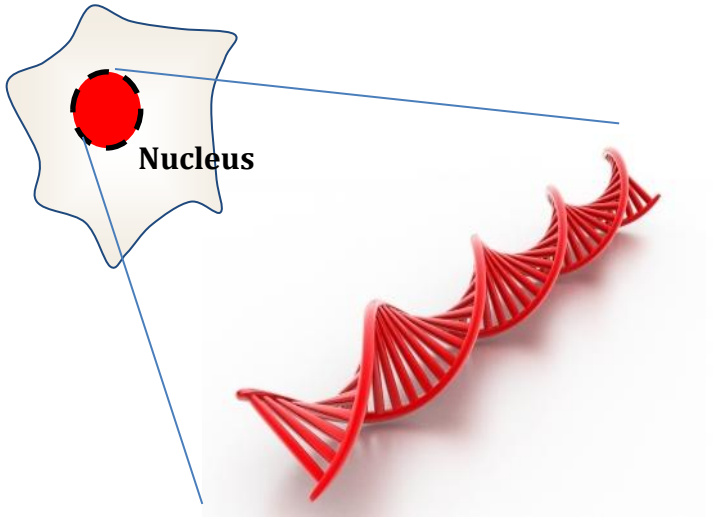
- ❑ Patients are few and geographically spread
- ❑ Research is fragmented
- ❑ Resources are limited
- ❑ Experts are few
- ❑ Specialised care centres for each disease cannot exist in every country
- ❑ Relevant information is little

RARE DISEASES ORIGINS

- Clinical manifestations
- Causes
- Populations they affect
- Severity and age of onset

The cell

80% Genetic origins



DNA (Deoxyribonucleic acid)

http://www.freedigitalphotos.net/images/view_photog.php?photogid=2888

- Bacterial or viral infections
- Allergies
- Environmental causes
- Degenerative or proliferative basis

Non-genetic origins

❑Suggestions that susceptibility may be genetically determined

DIAGNOSIS OF RARE DISEASES

Skin	Inverted nipples, abnormal subcutaneous fat pads at birth; fat pads disappear with increasing age
Neurologic	Global developmental delay; axial hypotonia, reduced deep-tendon reflexes, cerebellar atrophy, epilepsy; joint contractures, most children do not walk without support; stroke-like episodes
Nutrition/ gastrointestinal	Feeding problems (need for tube feeding common), gastroesophageal reflux, failure to thrive, hepatitis, liver failure
Ophthalmologic	Strabismus, impaired night vision, retinitis pigmentosa
Cardiac	Hypertrophic cardiomyopathy, pericardial effusion
Renal	Nephrotic syndrome early in life, renal cysts
Hematologic	Coagulation disorder (thrombosis or hemorrhage)

Box 2: Differential diagnosis of global developmental delay

- Infection (congenital)
- Trauma
- Exposure to toxins
- Uncontrolled seizures
- Asphyxia or hypoxic ischemic encephalopathy
- Structural brain abnormality
- Genetic, syndromic or neurocutaneous disorder
- Inborn error of metabolism
- Endocrinopathy
- Severe environmental deprivation or neglect
- Malnutrition or failure to thrive

Underrecognized

- ❑ **Several Specialists**
- ❑ **Several medical exams**
- ❑ **Delays in diagnosis**

BARRIERS IN ACCESS TO MEDICAL AND SOCIAL SERVICES

OUTLINE

- INTRODUCTION ABOUT RARE DISEASES (RD)
- THE CDG PATIENT'S VOICE
- LIVING WITH CDG

The CDG Patient's Voice

Links from different associations:

Portugal: http://sindromecdg.orgfree.com/
Spain: http://webs.ono.com/aescdg/SINDROME_CDG/Bienvenidos.html
France: http://www.lesptitscdg.org/
USA: http://www.cdgfamilynetwork.org/
Canada: http://www.thefog.ca/
http://thelittlefightersfoundation.com/
Germany: https://www.cdg-syndrom.de/
Denmark: http://www.cdgforeningen.dk/
Sweden: http://www.cdgs.se/

CDG active patient representatives nucleus:

Netherlands, UK, Ecuador, Brazil, Finland, Australia

The CDG Patient's Voice aims to:

Improve and **enhance understanding** and **awareness** of CDG
amongst the general public and medical professionals

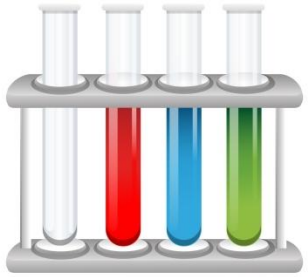
Facilitate tools for Patient Advocacy at International , national & EU levels ;

Support, promote and disseminate information to affected CDG families

FINAL GOAL:

TO INCENTIVE AND SUPPORT RESEARCH

http://www.freedigitalphotos.net/images/view_photog.php?photogid=1152

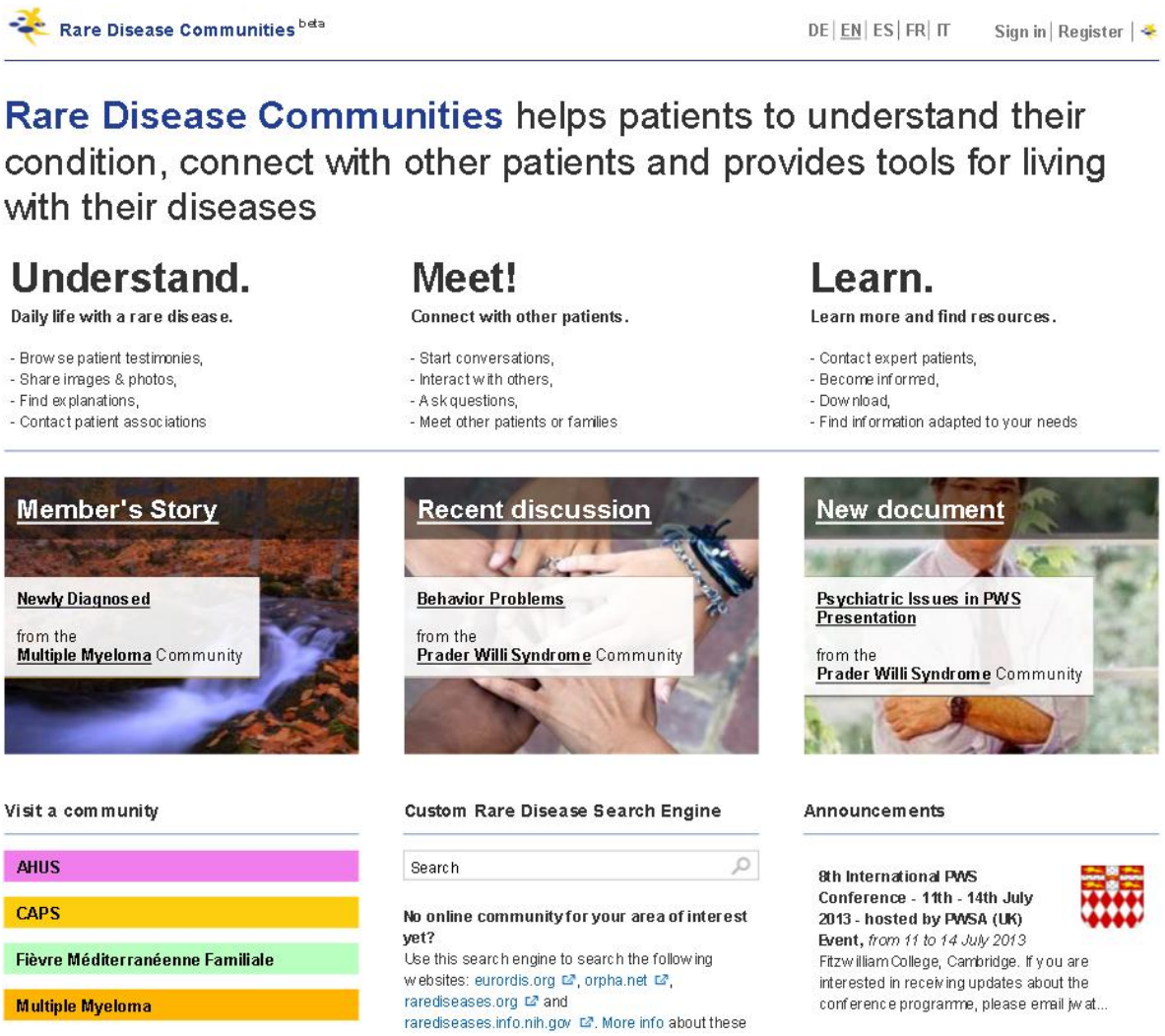


ww.freedigitalphotos.net/images/view_photog.php?photogid=2280

http://www.freedigitalphotos.net/images/view_photog.php?photogid=151

TO ESTABLISH A CDG NETWORK: PATIENTS, RESEARCHERS AND MEDICAL DOCTORS=RESULT!

Examples of CDG patient organizations initiatives:



http://www.rarediseasecommunities.org/en/community/cdg

Examples of CDG patient organizations initiatives:

- **Scientific ,medical and family Meetings**

I Luso-Hispanic Meeting for Congenital Disorders of Glycosylation
Barcelona , 21-22 of October 2011

Day 21 October
20.00: Welcome dinner

Day 22 October
8.30-9.00: Bring kids to kindergarten (The volunteer service is kindly offered by Cruz Roja)
9.00-11.00: **"CDG Syndrome: Past, Present and Future"**
Conference by
Dr Jaak Jaeken (Pediatric Department and Professor at the Faculty of Medicine at the University of Leuven, Belgium)
Dr Belén Pérez-Dueñas (Neurology department, Hospital Sant Joan de Déu, Barcelona, Spain)
Scientific collaborator: Dr Rafael Artuch (Clinical-Biochemistry Department, Hospital Sant Joan de Déu, Barcelona
11.00-11.30: Coffee
11.30-13.00: **"The Hispano-Luso-Argentina landscape about CDG Syndrome".**
Round table with:
Dr Mercedes Pineda (Neuropediatrician department,Hospital Sant Joan de Déu, Barcelona, España)
Dr Dulce Quelhaz (Genetic biochemistry Unity, Instituto de Genética Jacinto Magalhaes, Portugal)
Dr Paz Briones (Molecular Genetics and Biochemistry Unity, Instituto de Bioquímica Clínica, España)
Dr Carlo Ateaginni (Researcher at Centro de Estudio de las Metabolopatías Congénitas, Facultad de Ciencias Médicas, Universidad Nacional de Córdoba (UNC) , Argentina)
Scientific collaborator: Dr Vilaseca (Clinical Biochemistry Unity, Hospital Sant Joan de Déu, Barcelona, España)
13.00-15.00: Lunch at Casa del Mar
15.00-16.00: **"The clinical manifestations of CDG Syndrome: Prevention and treatment. Part I**
Round table with:
Dra. Ruth García Romero (Gastroenterology, Hepathology and child nutrition department, Metabolic Diseases Unity, Hospital Sant Joan de Déu, Barcelona)
Dr. Luiz Terricabras Coroli (Orthopedic department at Hospital Sant Joan de Déu, Barcelona)
Scientific collaborator: Vanessa Ferreira (Portuguese and Spanish CDG Syndrome Associations)
16.00-17.30: **"The clinical manifestations of CDG Syndrome: Prevention and treatment. Therapies. Part II.**
Round table with:
Dr Belén Pérez (Genetic diagnosis of inherited metabolic disorders Department, Universidad Autónoma de Madrid, España)
Dr Jaak Jaeken (Pediatric Department and Professor at the Faculty of Medicine at the University of Leuven, Belgium)
Dr Belén Pérez-Dueñas (Neurology department, Hospital Sant Joan de Déu, Barcelona, Spain)
Scientific collaborator: Dra Giró (Molecular Genetics and Biochemistry Unity , Instituto de Bioquímica Clínica, España)
17.30-18.00: Coffee
18.00-20.00: **"CDG Syndrome Brainstorming with families, Researchers and Medical Doctors"**: fundraising, practical guide for CDG families, future research projects, establishment of collaborations between the key stakeholders, etc..
20.15: Dinner at "La Casa del Mar"

Meeting directed to CDG Syndrome Medical doctors, Researchers and families!

Organizers:

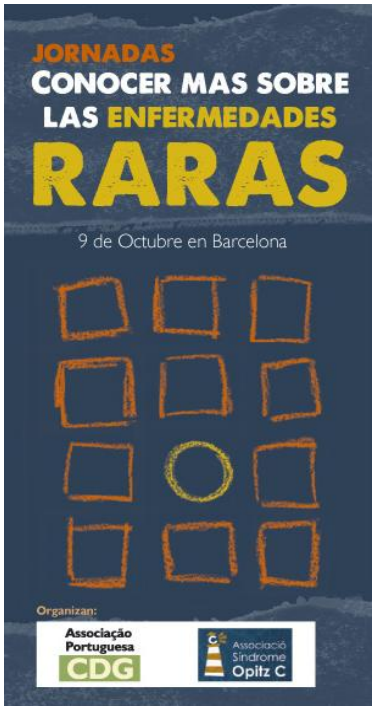
Collaborators:

Where: Casa del Mar
Calle Albareda, 1-13
Barcelona

Contact:
Vanessa Ferreira
sindromecdg@gmail.com

Note: The conference will be mainly in Spanish and Portuguese (although the slides will be in english).

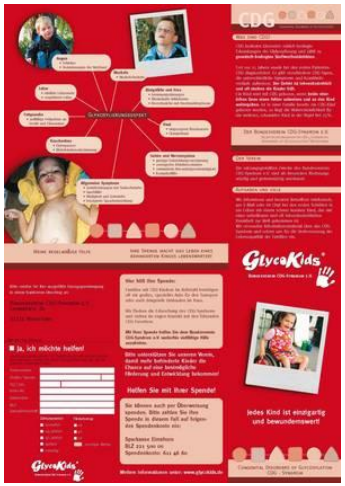
Glycokids CDG –
Family-Meeting 2011

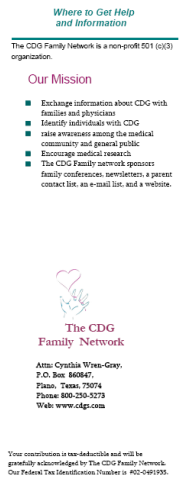


Examples of CDG patient organizations initiatives:

❑ Information/Dissemination for families, Medical Doctors and Researchers:

- Newsletter
- Access to scientific articles
- Leaflets
- Webpage
- Practical guide for CDG families





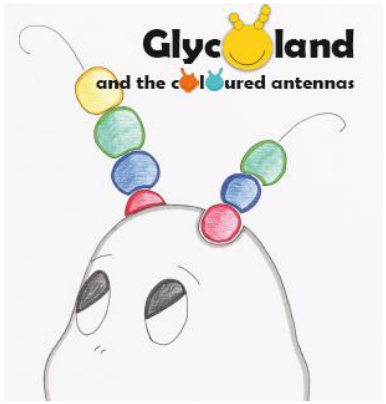
❑Awareness

- **High School and Universities projects**
- **CDG Community Store**

http://www.cafepress.com/CDG_Community

- **Fairy-tale about CDG:**

Glycoland and the coloured antennas



❑Understanding

Families give seminars:
Having a rare disease

OUTLINE

- INTRODUCTION ABOUT RARE DISEASES (RD)
- THE CDG PATIENT'S VOICE
- LIVING WITH CDG

Having a rare disease:

LIVING WITH CDG

29 month 200?

15 november 200?

- You are entering a ‘new’ strange world…
- … with ‘1001’ questions,
- But no answers what so ever!
- And the world outside keeps on going, and going and…

Finally at home..!! *Although..?!?!*

*"…Also the feeling of **uncertainty** is coming back. **Aspecially** now that – name of the child- is sleeping in his bed for the first time. **No monitor** just to check once a while, no sister who can call a doctor. **No doctor** that checks your child. Learing to trust on what you see, learning to trust on what your hear, learing to trust your child… Just **learning to trust** on your **own intuintion** makes it even more complicated"*

24-7

Non stop...

- From living to ‘survival’

Structuring & Organizing

24-7

Non stop...

‘Our 24 hour care language’

Visiting doctors	1 hour
‘Happiness’ hour	1
Administration	1 hour
Houshold	2 hours
Patients tools	2 hours
Medication hours	3
Therapies	7 hours
Taking rest & sleep	12 hours

“What did you miss??”

‘Questions & answers’ don’t match!!

supply and demand do not exclude each other

Protocols
Rules
Management
Laws
Agreements
Regulations etc…

knowledge and experience is up for grabs…

Knowledge and experience of

Parents

Thanks to:

- Families and members
- **We are grateful to …..**

You can mention research and medical groups from your country that are involved in CDG research

Sources of photos:

- Slide 5: from the left to the right
  - http://www.freedigitalphotos.net/images/view_photog.php?photogid=1499
  - http://www.freedigitalphotos.net/images/view_photog.php?photogid=345
  - http://www.freedigitalphotos.net/images/view_photog.php?photogid=809
  - http://www.freedigitalphotos.net/images/view_photog.php?photogid=1962
  - http://www.freedigitalphotos.net/images/view_photog.php?photogid=739
  - http://www.freedigitalphotos.net/images/view_photog.php?photogid=2741
  - http://www.freedigitalphotos.net/images/view_photog.php?photogid=2280
  - http://www.freedigitalphotos.net/images/view_photog.php?photogid=2367

- Slide 20: http://www.freedigitalphotos.net/images/view_photog.php?photogid=2125
- Slide 21

http://www.freedigitalphotos.net/images/view_photog.php?photogid=3062

- Slide 22

http://www.freedigitalphotos.net/images/view_photog.php?photogid=989

- Slide 23

http://www.freedigitalphotos.net/images/view_photog.php?photogid=987

- Slide 28

http://www.freedigitalphotos.net/images/view_photog.php?photogid=3062
http://www.freedigitalphotos.net/images/view_photog.php?photogid=3062

- Slide 29

http://www.freedigitalphotos.net/images/view_photog.php?photogid=3062

- Slide 30

http://www.freedigitalphotos.net/images/view_photog.php?photogid=2664

Before starting the awareness and dissemination talks

- Think on the targeted places where you plan to do the awareness and dissemination activities
- Send them a letter explaining who you are and the goals that you want to achieve;
- Inform us about the GREAT results that you got with your activity by sending an e-mail to **cdgawareness@gmail.com**

CDG AWARENESS AND DISSEMINATION KIT :

- Kit materials include:

  - ❑A powerpoint with an introduction about Rare Diseases and Living with a Rare Disease (CDG in our case)

  - ❑Fundraising and Conference presentation course

For CDG AWARENESS AND DISSEMINATION KIT materials, please contact:
cdgawareness@gmail.com

CONGENITAL DISORDERS OF GLYCOSYLATION (CDG) AWARENESS AND DISSEMINATION PROJECT

QUESTIONS ABOUT PRESENTATION SUPPORT CONCERNING:

- **INTRODUCTION ABOUT RARE DISEASES (RD)**
- **THE PATIENT'S VOICE**
- **CONFERENCE PRESENTATIONS COURSE**

CONTACT: Vanessa Ferreira (sindromecdg@gmail.com)

- **HAVING A RARE DISEASE: LIVING WITH CDG**

CONTACT: Bas Holten (basesdownunder2003@hotmail.com)

TUTORIAL FOR FUNDRAISING:

- **SIMPLE STEPS FOR FUNDRAISING SUCCESS**

CONTACT: Andrea Berarducci (maui911@yahoo.com)

CONGENITAL DISORDERS OF GLYCOSYLATION (CDG) AWARENESS AND DISSEMINATION PROJECT

In each of the following slides, we suggest some sources of information and ideas about what you may want to discuss and highlight in your presentation…

Be aware that some information should be adapted to the country where you live. Thanks!

SKYPE FOR HELP IN ORAL PRESENTATION PREPARATION:
Skype name: cdgawareness

PLEASE IF YOU USE THIS AWARENESS MATERIAL WE KINDLY REQUEST YOU TO MENTION AS SOURCE:

CDG AWARENESS DONE BY CDG PATIENT'S VOICE

TUTORIAL FOR FUNDRAISING:

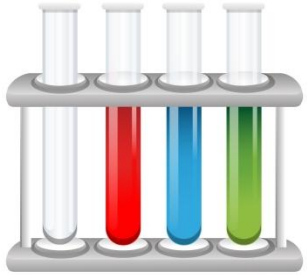
- **SIMPLE STEPS FOR FUNDRAISING SUCCESS**

CONTACT: Andrea Berarducci (maui911@yahoo.com)

CDG PATIENT'S VOICE AIMS:

TO INCENTIVE AND SUPPORT RESEARCH

http://www.freedigitalphotos.net/images/view_photog.php?photogid=1152



ww.freedigitalphotos.net/images/view_photog.php?photogid=2280

http://www.freedigitalphotos.net/images/view_photog.php?photogid=151

TO ESTABLISH A CDG NETWORK: PATIENTS, RESEARCHERS AND MEDICAL DOCTORS=RESULT!

Simple Steps TO Fundraising success

Sources of Funding - Overview

- Individuals
- Foundations – Large, Community, Corporate
- Corporations – Large, Community-based
- Government

Fundraising Basics

- Set a goal
  - Identify the need
  - Be clear about what the money will be used for
- Target an audience
  - Know or learn who your audience is
  - What will attract your audience to the cause?
- Formulate a plan
  - How much money do you want to raise?
  - Who will be donating toward the cause?
  - Who can help you put this plan/event together?
  - What is a realistic time frame?

Fundraising Options

- Auctions
- Selling merchandise / services
- Themed breakfasts / dinners
- Run/Walk/Bike – athons
- Events with a twist… comedy night, trivia night, bowling events, dance-a-thons
- The “Non-event” event
  - A mailing or solicitation in place of an actual event
  - Cash cans, percentages of breakfast / dinner sales, etc

Selecting the “Right” Fundraiser

- Group dynamics
  - Research your target audience
  - “Think globally, act locally”
- Establish a time frame
  - Consider time, money and resources available
- Consider costs
  - Network! Look for those who may be willing to donate goods and/or services toward your fundraiser
- Identify resources
  - What materials are needed? What manpower is needed?
  - Spread tasks among volunteers according to experience, strengths and capabilities

The Details of the Fundraiser

- Identify a location – look for donated space, work with the venue coordinator
- Setting the "right" date – prepare for "seasons" and weather considerations, if applicable
- Organizing and recordkeeping – be accountable, track donors, and collect contact information for follow-up!
- Finding volunteers – who and how can people help?
  - Churches, civic groups, schools, etc
- Set a schedule
  - Be comprehensive, include details, track progress
- Invitees and special guests
  - Think of anyone who will draw the audience to the cause
- Contingency plans – always have a back-up plan

Community Fundraising

- Involving the community – focus on everyone, not just the “big” players in the community
- Make things happen – start small, get people on board, and build group enthusiasm
- Work with your community Board – build awareness
- Find local sponsors – find sponsors, co-sponsors and make opportunities for recognition
- Promote locally – pre-event coverage as well as post-event coverage; give yourself at least 3 months to promote your fundraiser
- Social networking – post event information, pictures, videos and blogs

The Budget

- Assess costs – “Raising money costs money”
  - Typical expenses – food/refreshments, speakers/equipment, travel/lodging, items to be sold/raffled, event supplies
- Budget for expenses – Determine how you will spend money and how you will make money
  - Sell tickets, hold raffles, auction items, etc
- Find sources of funding
  - Develop a list of prospects
  - Individuals, businesses, civic organizations, government agencies, foundations and trade associations

Fundraising Tools

- Communication infrastructure
  - Determine how technically savvy your group is
  - Identify technology available to the group, as well as budget or equipment that will be necessary for your communication strategy
- Know who to contact
  - Make a list of contact persons for each aspect of planning
- Using a website for fundraising
  - Post pictures, videos, interviews and blogs
  - Social networking, email and facebook
- E-fundraising
  - Low cost way to generate revenue in support of a specific cause
  - Causes.com, Justgive.org, etc

Spreading the Word

- Advertising – Define your advertising needs:
  - Radio – PSAs, on-air interviews/appearances
  - Internet & Email – post stories, pictures & videos
  - Online newsletters – always link back to your website
  - Signs, Flyers and Posters – simple, clear and strategic placement
  - Promotional Activities – free giveaways, kickoff event, photo opportunities and print stories prior to the event
  - Print – use community resources, newspapers, grocery stores, community centers, etc
  - Visual Marketing – "A picture is worth a thousand words"
    - Design the graphics, text and photos for your event in a visually appealing and clear way
    - Hire or recruit a professional to donate services to help

Alternative Ways of Fundraising

- Foundations
- Corporations
- Grants
- Other major donors

Foundations

- Can be public, private, corporate or government foundations
- Typically fund worthwhile causes and activities including educational, scientific, environmental, political and charitable needs

- Foundations are typical funders of special projects and do not fund ongoing operating expenses
- Seeking foundation grants require well established plans and submission of a proposal, including measuring evaluation and follow-up

- Research the Foundation Center – www.fndcenter.org

Corporations

- Approaching Corporations
  - Who do you know?
  - Build relationships
  - Give something back – public image, corporate recognition

- Employee Donations
  - Many companies have employee contribution programs or “matches”
  - Contact the company’s Human Resources and to discuss matching gift programs (either adding to or establishing a new one for your cause)

- The United Way – currently contributes to 1,300 community-based organizations

Grants

- Finding grants
  - Do your research, be aware of geographical restrictions, guidelines of the funder, application criteria and whether your needs are a match for the funding programs available
- The application process
  - Collect data, be specific, state your cause, following grant guidelines, develop collaborations and support, prepare the presentation
- Timing and follow-up
  - Grant funding is competitive, prepare reasonable time-lines

- Corporate grants – make sure that your cause if a good match for the corporation
- Federal grants – search for grants applicable to your cause; www.grants.gov

Measuring Success

- Evaluate fundraising efforts – post-event evaluations… “what worked” and “what needs improvement”
- Evaluate the process – did you make the right choices, what are areas that you can measure for future success
- Make improvements – seek feedback, judge your weak points, focus on the positives
- Fundraising wrap-up – maintain a log of resources and event highlights, prepare for the future, report results
- Say “thank you”, “thank you”, “thank you” to everyone who helped make your event successful, ensure that proper acknowledgements are made for “repeat performances”!

Conference presentation Course

http://www.freedigitalphotos.net/images/view_photog.php?photogid=1738

- **IDEAS TO REACH A GREAT TALK**

Tutorial by Vanessa Ferreira (sindromecdg@gmail.com)

Points to consider:

- Effective public Speaking
- Anatomy of a seminar
- Common mistakes in slide preparation

Effective public Speaking

- Voice Projection

(Speak loud and clear, but dont shout!)

- Make eye contact with your audience

- Be animated and enthusiastic about your work

To avoid this:

http://www.freedigitalphotos.net/images/view_photog.php?photogid=1499

How Do I Motivate Others?

- Verbalize an Inspiring Vision and Mission
- Project a Courageous Spirit
- Behave with Enthusiasm

Anatomy of a seminar

- It’s mportant to structure your content!

- Structure helps memory and orientation.

http://www.freedigitalphotos.net/images/view_photog.php?photogid=659

Anatomy of a talk

It’s like a sandwich!

http://www.freedigitalphotos.net/images/view_photog.php?photogid=2365

INTRODUCTION

BODY OF THE TALK

- **E**VIDENCE
- **I**NFORMATION
- **A**RGUMENT

CONCLUSION

INTRODUCTION

-Name / Role

-Purpose of Conference

-Agenda

-Elucidate the public if they can interrupt you during the talk or if the questions should be at end

INTRODUCTION

- **Your purpose and/message**
  - What will they learn once they leave your talk that they do not have now?
- **Importance of the message**
  - Why should they bother?
- **Orientation**
  - Starting point, what will be covered, where end?

BODY OF THE TALK

To organise your:

- **E**VIDENCE
- **I**NFORMATION
- **A**RGUMENT

Into a logical flow.

CONCLUSION

- State message (again)
- Summarise
- Why it is important
- Thank them (motivate the audience to questions!)

SECRET IS….

PLAN

PLAN

PLAN

PRACTICE

PRACTICE

PRACTICE

Common mistakes in slide preparation

- Time: 1 minute per slide! And do not prepare long talk (25-30 minutes)
- Choice of font and point size
- Choice of text color
- Improper use of animation

Choice of font and point size

- 24 point font use for text
- 32 point font use for list of points
- 48 point font use for titles

Choice of text color

- In fact the option to use color is very subjective. Although, it is important to be aware about the combination of colours that you use:
  - Blue and black it is is a terrible combination, like yellow and white

Then, we cannot forget that 7% of male population is red/green blind

Improper use of animation

- Use the animation only when required, otherwise it can distract.

- And make sure that you trained the use of the animation .

GOOD LUCK!!!!!! AND….

http://www.freedigitalphotos.net/images/view_photog.php?photogid=2734

Bibliography:

Smith R. How not to give a presentation. *British Medical Journal, 2000, 321:1570*–1571.

http://www.weizmann.ac.il/mcb/UriAlon/nurturing/HowToGiveAGoodTalk.pdf