D7.3 “Project leaflet for individuals with DS, their families, clinicians and scientists and professional templates”

WP7 – “Dissemination, communication, exploitation and guideline development”

GO-DS21

GENE OVERDOSAGE AND COMORBIDITIES DURING THE EARLY LIFETIME IN DOWN SYNDROME

Disclaimer

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Copyright message

Document information

Grant Agreement Number	**848077**	**Acronym**	**GO-DS21**
Full title	Gene Overdosage and Comorbidities During the Early Lifetime in Down Syndrome		
Topic	Understanding causative mechanisms in co- and multimorbidities combining mental and non-mental disorders		
Funding scheme	RIA - Research and Innovation action		
Start Date	1 January 2020	**Duration**	60 months
Project URL	www.go-ds21.eu		
EU Project Officer	Monica Ensini		
Project Coordinator	Yann Herault, CENTRE EUROPEEN DE RECHERCHE EN BIOLOGIE ET MEDECINE (CERBM)		
Deliverable	D7.3		
Work Package	WP7		
Date of Delivery	**Contractual**	30th Sep 2020	**Actual**	23rd Sep 2020
Nature	Other	**Dissemination Level**	Public
Lead Beneficiary	3-CRG		
Responsible Author(s)	Andrea Wohner and Nina Donner (concentris)		
Keywords	Project leaflet, project presentation slides, letterhead		

History of changes

Version	Date	Contributions	Contributors (name and institution)
1.0	January 2020	Development of GO-DS21 letterhead template	Andrea Wohner, Elmar Schwalber (08 concentris) Yann Herault (01 CERBM)
1.0	January 2020	Development of GO-DS21 powerpoint template	Andrea Wohner, Nina Donner (08 concentris)
1.0	September 2020	Development of GO-DS21 project slides	Yann Herault (01 CERBM), Andrea Wohner (08 concentris), 06 Johannes Beckers (HMGU)
1.0	September 2020	Development of GO-DS21 project leaflet (English version)	Nina Donner, Andrea Wohner (08 concentris), Yann Herault (01 CERBM), André Strydom (04 KCL, 11 T21RS), Mara Dierssen (03 CRG), Johannes Beckers (06 HMGU), Pat Clarke (EDSA)
1.0	September 2020	Development of GO-DS21 project leaflet (French version)	Sophie Durand, Alix de Cleene (07 IJL), Yann Herault (01 CERBM), Andrea Wohner (08 concentris)
1.0	September 2020	Development of GO-DS21 project leaflet (Spanish version)	Rafael De La Torre, Maria Gomis González, Laura Forcano Gamazo (09 IMIM)

1 Objectives of the deliverables based on the Description of Action

The project leaflet for individuals with DS, their families, clinicians and scientists, the letterhead template and powerpoint template serve the following WP7 objectives and tasks:

- **Objective 1**: To disseminate the results of GO-DS21 to relevant stakeholders
  - **Task 1**: Logo, website and corporate identity of GO-DS21 (concentris, CERBM, CRG, EDSA, T21RS)
  - **Task 3**: Communication to the public and to patient communities in Europe and beyond (EDSA, CRG, concentris, T21RS)
- **Objective 2**: To disseminate the results to the scientific, healthcare, pharmaceutical and policy sectors and foster interaction and exchange with DS organizations
  - **Task 2**: Dissemination for scientists and medical community (CRG, CERBM, concentris, T21RS)

2 Executive Summary / Abstract

This deliverable report contains:

1. GO-DS21 letterhead template (Annex 1)
2. GO-DS21 powerpoint template (Annex 2)
3. GO-DS21 project slides (Annex 3)
4. GO-DS21 project leaflet (Annex 4 – English version; Annex 5 – French version; Annex 6 – Spanish version)

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 848077

3 Results

3.1 GO-DS21 letterhead template

A professional letterhead template has been designed by concentris that can be used by all GO-DS21 partners for project related communication with relevant stakeholders. The template can be downloaded from the password-protected GO-DS21 intranet (see Annex 1).

3.2 GO-DS21 powerpoint template

A set of GO-DS21 PowerPoint slides has been developed (see Annex 2); these slides are used by all partners for presentations at GO-DS21 internal meetings, conferences and other presentation opportunities (see Annex 2). The slides are designed in the corporate design of GO-DS21 to contribute to the growing awareness of GO-DS21 and its EC funding.

3.3 GO-DS21 project slide corporate desk

Professionally designed presentation slides explaining the most important facts of GO-DS21 for experts, people with DS, their relatives and caregivers were developed by concentris. This presentation is available to all partners of GO-DS21 via the password-protected intranet and will be adapted throughout the entire project period in light of new findings (see Annex 3).

3.4 GO-DS21 project leaflet

In order to inform individuals with DS, their families and caregivers, clinicians, scientists and the general public about the goals of GO-DS21, concentris has designed a project leaflet in cooperation with the GO-DS21 partners 01 CERBM, 03 CRG, 04 KCL, 06 HMGU, 11 T21RS and EDSA. A digital version (web version) of the project brochure can be found on the project website (https://go-ds21.eu/news-events/downloads/). Printed versions of the project leaflet were sent to all GO-DS21 partners for distribution at conferences, in their institutions or clinics, and on other occasions (see Annex 4).

The project leaflet has also been translated into French with the help of 01 CERBM and 08 IJL (see Annex 5) and into Spanish with the help of 09 IMIM (see Annex 6). Printed copies of the French and Spanish versions have been sent to the French and Spanish partners. Additional copies or virtual ones will be proposed to our Third Party “European Down Syndrome Association” (EDSA) for dissemination within their network.

This project has received funding from the European Union’s Horizon 2020 research and innovation programme under grant agreement No 848077

Annex 1:
GO-DS21 letterhead template

INSTITUT DE GÉNÉTIQUE BIOLOGIE MOLÉCULAIRE ET CELLULAIRE – IGBMC
UNISTRA, CNRS, INSERM | 1 RUE LAURENT FRIES | 67404 ILLKIRCH CEDEX 2

Project management

concentris research management gmbh
Fürstenfeldbruck, Germany
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Scientific coordinator

Dr. Yann Hérault, PhD
INSTITUT DE GÉNÉTIQUE BIOLOGIE MOLÉCULAIRE ET CELLULAIRE – IGBMC
Illkirch, France

info@go-ds21.eu
www.go-ds21.eu

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 848077.

Annex 2:
GO-DS21 powerpoint template

GO-DS21 (Title)

Gene Overdosage and Comorbidities During the Early Lifetime in Down Syndrome (Subtitle)

John Doe (MD, PhD)
Name of partner institution

Strasbourg, France, 06 February 2020

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 848077.

Placeholder for a headline.

Insert content here …

A bullet list:

- Lorem ipsum dolor sit amet, consectetuer adipiscing elit
  - Aenean ligula eget dolor cum sociis natoque penatibus
- Et magnis dis parturient montes

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 848077.

GO-DS21 colour palette

Light blue
0 R 180 G 220B

Dark blue
0 R 50 G 110 B

Green yellow
220 R 220 G 20 B

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GO-DS21 table layout

Table Header		
Body content		

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 848077.

Blank page for images and/or illustrations

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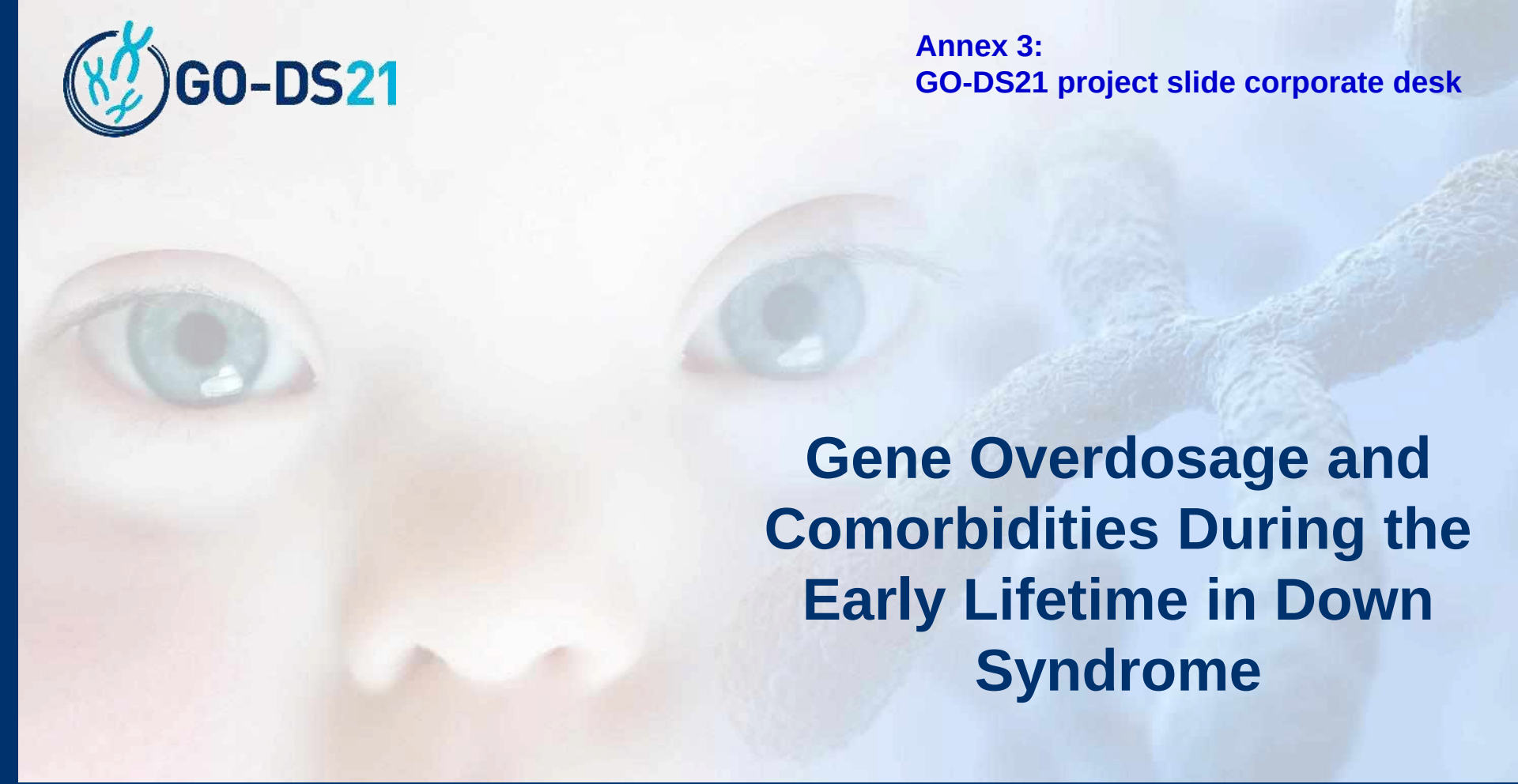
GO-DS21

Annex 3:
GO-DS21 project slide corporate desk



Gene Overdosage and Comorbidities During the Early Lifetime in Down Syndrome

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 848077.

GO-DS21 IN A NUTSHELL

Full project title	**Gene Overdosage and comorbidities during the early lifetime in Down Syndrome**
Start date	**1st January 2020**
Duration	**60 months (5 years)**
Participants	**12 institutions and 1 patient organization from 6 European countries**
EC funding	**~ 6 million €**
Project website	**go-ds21.eu**

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 848077.

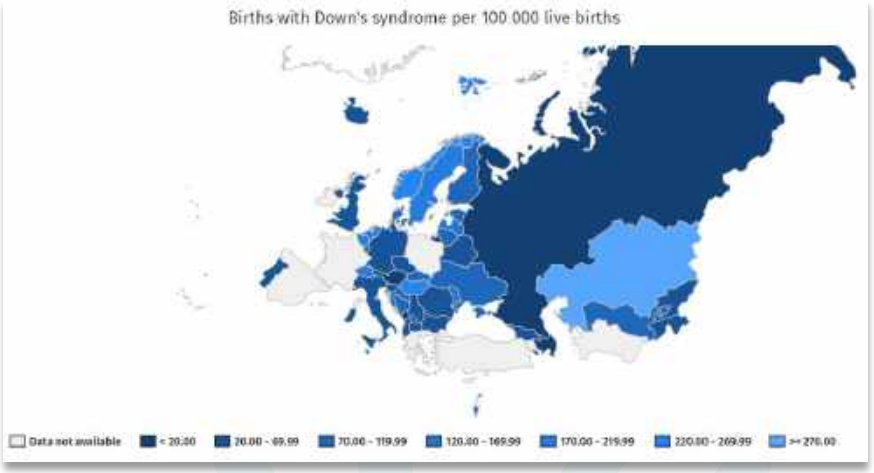
RESEARCH CONSORTIUM

01	Centre Européen de Recherche en Biologie et Médecine (CERBM, France): Dr Yann Herault
02	Institut du Cerveau et de la Moëlle Épinière (ICM, France): Dr Marie-Claude Potier
03	Centre for Genomic Regulation (CRG, Spain): Dr Mara Dierssen
04	King's College London (KCL, UK): Prof Andre Strydom
05	Queen Mary University of London (QMUL, UK): Prof Li Chan
06	Helmholtz Zentrum Munich – German Research Center for Environmental Health (HMGU, Germany): Prof Johannes Beckers
07	Institut Jérôme Lejeune (IJL, France): Dr Sophie Durand
08	concentris research management GmbH (concentris, Germany): Ms Andrea Wohner
09	Fundació Institut Mar d'Investigacions Mèdiques (IMIM, Spain): Prof Rafael De La Torre
10	The Chancellor Masters and Scholars of the University of Cambridge (UCAM, UK): Prof Pietro Lio'
11	Trisomy 21 Research Society (T21RS, Netherlands): Prof Andre Strydom
12	Perha Pharmaceuticals (PERHA, France): Dr Laurent Meijer
13	European Down Syndrome Association (EDSA, Belgium): Mr Pat Clarke

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 848077.

WHY GO-DS21 MATTERS – Meet the „unmet“ need

- **Down syndrome (DS)** is the **most common genetic form** of **intellectual disability (ID)** in the world, with an incident of 1 in 1,000 births, affecting **more than 5 million people worldwide**.
- Mental and physical conditions, such as **intellectual disability and obesity**, appear at a much **higher rate in individuals with Down syndrome** compared to the general population.
- The overarching aim of GO-DS21 **is to elucidate etiological mechanisms involved in the common morbidities of obesity and intellectual disability.**
- More precisely, GO-DS21 will **explore how intrinsic (genetic and pathway-driven) and extrinsic (environmental and epigenetic) factors can lead to such comorbid states.**



Number of births with DS per 100 000 live births (WHO European Health Information Gateway, 2012 – interactive map)

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 848077.

GO-DS21 RESEARCH OBJECTIVES

Objective 1: Determine **comorbidity patterns** and associated factors seen during the **early lifetime** in **persons with Down syndrome**

Objective 2: Decipher the **contribution of environmental factors** to **Down syndrome comorbidities**

Objective 3: Investigate the **effects of overdosage** of three **Hsa21 candidate driver genes** to explain comorbid patterns in Down syndrome

Objective 4: Identify **specific physiological biomarkers** and **regulatory and epigenetic signatures** derived from human samples and rodent models

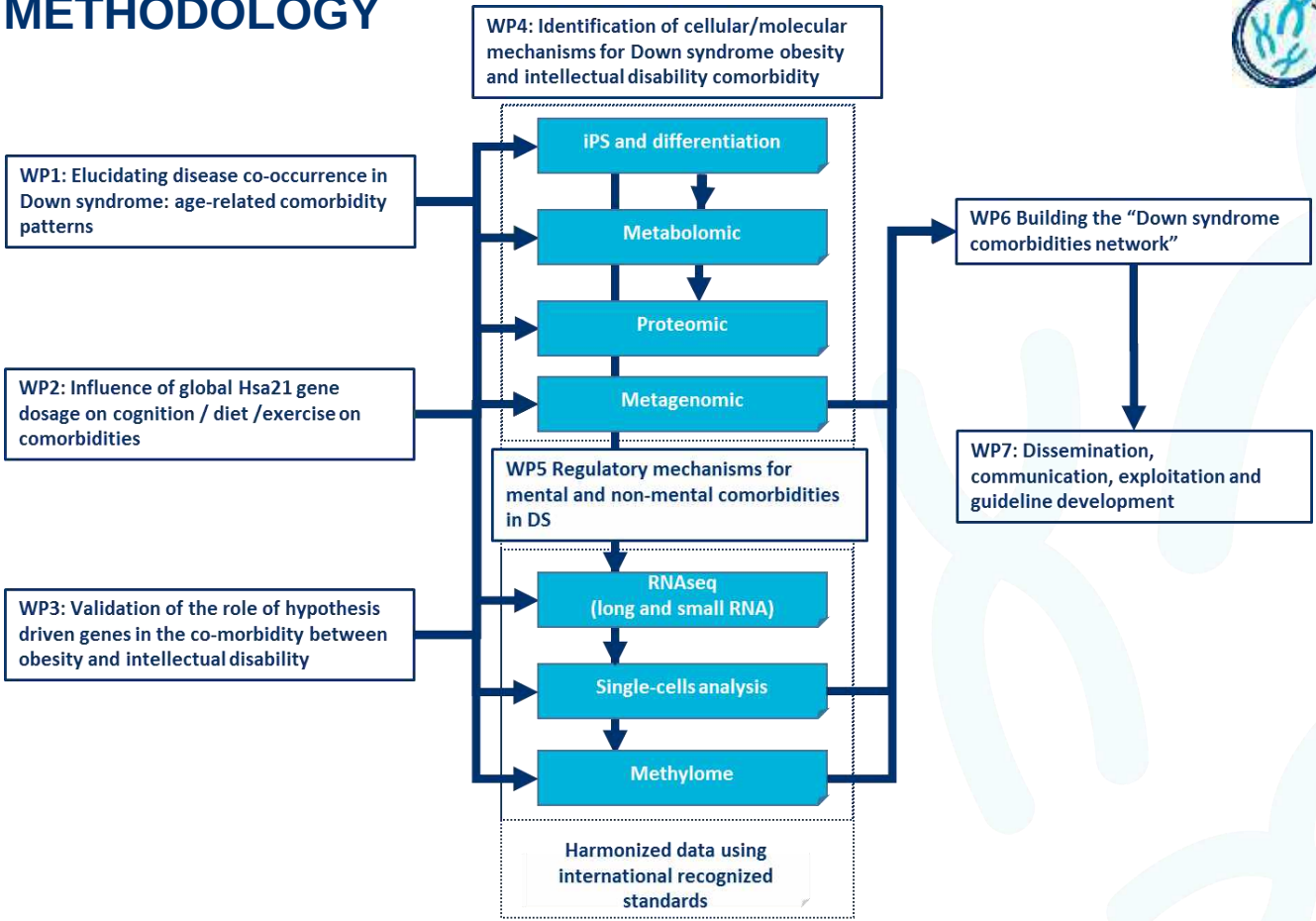
Objective 5: **Integrate various types of data** from human samples and preclinical models across different spatial and temporal scales of biological complexity using **computational biology models** and **machine learning approaches**

Objective 6: Design **new therapeutic interventions with pharmacological compounds**, **gene therapy** approaches and **environmental intervention** to **reduce the penetrance of comorbidities in preclinical models**

Objective 7: Based on the new findings in GO-DS21, **new international recommendations** and **targeted interventions** to **prevent or minimise the appearance of obesity** within the context of intellectual disability in people with Down syndrome will be established and disseminated to **all relevant stakeholders**

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 848077.

GO-DS21 METHODOLOGY



This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 848077.

MAIN INNOVATIONS AND EXPECTED IMPACT OF GO-DS21

A new paradigm for understanding gene overdosage in comorbidities associated with DS, particularly ID and obesity.
Specifically, we will:

1) **Elucidate the role of genetic risk** of these and associated **comorbidities,** focusing on **Hsa21 genes** that may be **relevant not only for DS, but also for other populations.**
2) Shed light on **stress-related mechanisms** in the development and maintenance of comorbid obesity and ID.
3) **Deliver novel experimental models** for comorbidities such as new mouse models and human cellular models.
4) **Identify biomarkers** for more accurate and earlier diagnosis and monitoring of comorbidities.
5) **Provide new opportunities to develop evidence-based innovative combined-therapy** for obesity/cognitive impairment.
6) **Provide clinically applicable user-adapted guidance** for the management of comorbidity by developing **two-way engagement** with stakeholders.

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 848077.

CONTACT

Scientific Coordination	**Dr. Yann Herault (IGBMC-CERBM)** Illkirch-Graffenstaden, France herault@igbmc.fr
Project Management	**Andrea Wohner (concentris)** concentris research management GmbH Fürstenfeldbruck, Germany andrea.wohner@concentris.de
Project website	**go-ds21.eu**
Follow us on Twitter	**twitter.com/GO_DS21**

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 848077.

Annex 4: GO-DS21 project leaflet - EN

RESEARCH CONSORTIUM

12 institutions and 1 patient organisation from 6 European countries with combined expertise in human and mouse phenotyping, molecular biology, and computer modelling

Scientific Coordination
Consortium Partner
Patient Organisation

GO-DS21 IN A NUTSHELL

Full project title	**G**ene **O**verdosage and comorbidities during the early lifetime in **D**own **S**yndrome
Start date	01 January 2020
Duration	60 months (5 years)
Participants	12 institutions from 6 European countries
EC funding	6 million €
Project website	go-ds21.eu

CONTACT

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Follow us on Twitter: twitter.com/GO_DS21

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No 848077.

Photo credits: DenKuvaiev, BlackJack3D

Elucidating the link between intellectual disabilty, obesity, and other comorbities

WHY IT MATTERS

Down syndrome is the most common genetic form of intellectual disability in the world, with an incidence of 1 in 1000 births, affecting more than 5 million people worldwide. It is characterized by an extra copy of human chromosome 21 (trisomy 21). Within the European population, the prevalence of Down syndrome remains high due to increased maternal age and lack of prenatal monitoring in certain at-risk populations. In addition, increased life expectancy makes people with Down syndrome a significant proportion of the population with a need to be better investigated in order to improve their health-care.

Mental and physical conditions, such as intellectual disability and obesity, appear at a much higher rate in individuals with Down syndrome compared to the general population. Obesity in people with Down syndrome also increases their risk of developing obstructive sleep apnea (a potentially deadly breathing disorder), dyslipidemia (an abnormal amount of lipids in the blood), hyperinsulinemia, type-2 diabetes, and gait problems.

Understanding the underlying causes and biological pathways for these conditions is therefore of major importance to improve the health of people with Down syndrome. Despite the complexity, the genetic cause of Down syndrome is clear: the genes on chromosome 21 are present in overdose (3 copies instead of 2). This will help us to unravel the underlying mechanisms for obesity and other comorbidities.

REAL-WORLD IMPACT

GO-DS21 uses newly generated preclinical and clinical data from existing clinical cohorts in addition to newly generated data. Data points emerge from the so-called "Down syndrome comorbidities network" which includes data from cohorts in the UK (more than 6,000 cases), France, Spain, and three other European clinics. Next to the clinical data sets, GO-DS21 will investigate project-relevant hypotheses in preclinical animal models for a better understanding of Down syndrome. We explore how intrinsic (genetic and pathway-driven) and extrinsic (environmental and epigenetic) factors lead to common comorbidities. The project's results may also be applicable to people suffering from other types of intellectual disability.

- → Novel biomarkers for accurate and early diagnosis
- → Innovative new therapies and novel national and international clinical guidance
- → Prevention and better management of obesity
- → Improved management of intellectual disabilities
- → Enhanced monitoring through clinical measurements and fluid biomarkers
- → Better understanding of the interaction between obesity and cognitive impairment
- → Increased visibility and awareness of Down syndrome and related research

Down syndrome is the most common genetic form of intellectual disability in the world

Photo credit: SDI Productions

Photo credit: Iuzsek

OUR VISION

GO-DS21 aims to identify and validate new causative mechanisms underlying comorbid conditions, primarily intellectual disability and obesity, in Down syndrome. People with Down syndrome often have a combination of mental conditions (intellectual disability, autism, or affective disorders) as well as physical impairments (cardiac and haematological defects, obesity, and gastrointestinal abnormalities).

On a larger scale, our research will not only increase the understanding of these comorbidities in Down syndrome but also elucidate the link between obesity and mental conditions in the general population. Obesity is a major risk factor for many chronic diseases, contributing to the death of more than 2.8 million people every year. Moreover, obesity is related to cognitive impairment, increasing dramatically in populations with intellectual disability.

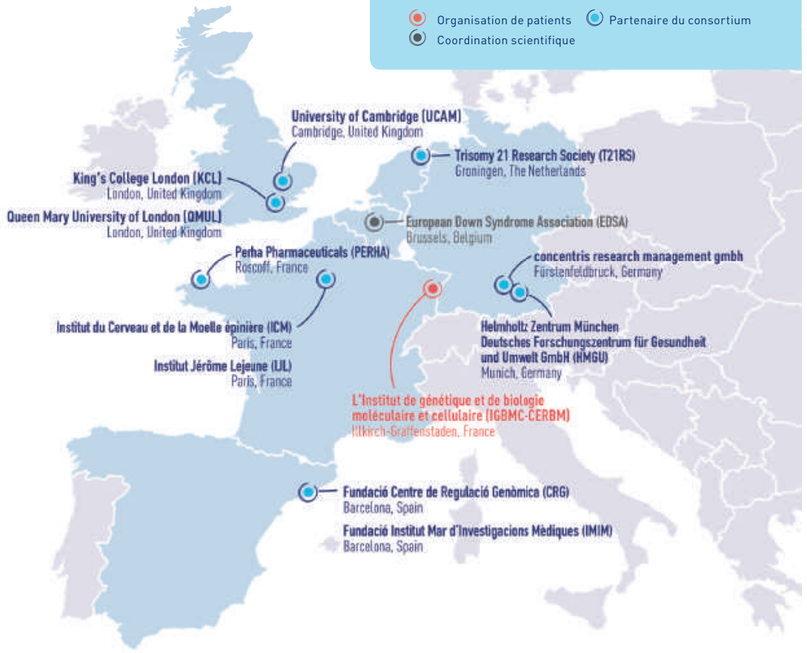
The innovative approach of this interdisciplinary research project unites basic and clinical researchers as well as experts on pathophysiology, bioinformatics, and artificial intelligence. GO-DS21 strives to improve early diagnosis, prognosis, and treatment of conditions that co-occur in Down syndrome. We will establish new clinical recommendations and identify novel and well-targeted early-lifetime interventions to prevent or at least minimize the development of obesity within the context of intellectual disability. These may also have application in the general population.

Annex 5: GO-DS21 project leaflet - FR

CONSORTIUM DE RECHERCHE

12 institutions et 1 organisation de patients de 6 pays avec une expertise combinée en analyse fonctionnelles (physiologie, cognition) humain et murin, en biologie moléculaire et en modélisation Informatique

Organisation de patients
Partenaire du consortium
Coordination scientifique



GO-DS21 EN QUELQUES MOTS

Titre complet du projet	Surdosage génétique et comorbidités au début de la vie dans la trisomie 21
Date de début	01 janvier 2020
Durée	60 mois (5 ans)
Participants	12 institutions et 1 organisation de patients de 6 pays européens
Financement de la CE	6 millions d'euros
Site web du projet	go-ds21.eu

CONTACT

Coordination scientifique
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Illkirch-Graffenstaden, France
herault@igbmc.fr

Gestion de projet
Andrea Wohner
concentris research management gmbh
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Suivez-nous sur Twitter: twitter.com/GO_DS21

Le présent projet a bénéficié d'un financement au titre du programme-cadre de l'Union européenne pour la recherche et l'innovation „Horizon 2020" dans le cadre de la convention de subvention n° 848077

Photo credits: DenKuvaiev, BlackJack3D

Elucider le lien entre déficience intellectuelle, obésité et autres comorbidités

POURQUOI CELA EST-IL IMPORTANT?

La trisomie 21 est la forme génétique de déficience intellectuelle la plus répandue dans le monde, avec une incidence de 1 naissance sur 1000, touchant plus de 5 millions de personnes. Elle se caractérise par une copie supplémentaire du chromosome 21 humain (trisomie 21). Au sein de la population européenne, la prévalence de la trisomie 21 reste élevée en raison de l'augmentation de l'âge maternel et du manque de suivi prénatal dans certaines populations. De plus, avec une espérance de vie plus grande, less personnes porteuses de trisomie 21 représentent une part de plus en plus importante de la population qu'il est nécessaire d'étudier afin d'améliorer leur prise en charge médicale.

Certaines conditions mentales et physiques, telles que la déficience intellectuelle et l'obésité, apparaissent à une fréquence beaucoup plus élevée chez les personnes avec une trisomie 21 par rapport à la population générale. L'obésité chez les personnes avec une trisomie 21 augmente également leur risque de développer une apnée obstructive du sommeil (un trouble respiratoire potentiellement mortel), une dyslipidémie (une quantité anormale de lipides dans le sang), une hyperinsulinémie, un diabète de type 2 et des problèmes de marche.

Malgré sa complexité, la cause génétique de la trisomie 21 est claire: les gènes du chromosome 21 sont présents en surdose (3 copies au lieu de 2). Aussi comprendre les causes sous-jacentes et les voies biologiques de ces affections est donc d'une importance majeure pour améliorer la santé des personnes porteuses de trisomie 21. Avec cette étude nous voulons démêler les mécanismes sous-jacents de l'obésité et d'autres comorbidités.

IMPACT MONDIAL REEL

GO-DS21 utilise des données précliniques et cliniques nouvellement générées de cohortes cliniques existantes en plus de nouvelles données générées. Les sources de données proviennent du „réseau de comorbidités de la trisomie 21" qui comprend des données de cohortes du Royaume-Uni (plus de 6 000 cas), de France, d'Espagne et de trois autres centres cliniques européens. Outre l'ensemble de ces données cliniques, GO-DS21 étudie des hypothèses pertinentes pour le projet à partir de modèles animaux précliniques pour une meilleure compréhension de la trisomie 21. Nous étudions comment des facteurs intrinsèques (génétiques et liés aux voies de transmission) et extrinsèques (environnementaux et épigénétiques) conduisent à des comorbidités communes. Les résultats du projet pourront également s'appliquer aux personnes avec d'autres types de déficience intellectuelle.

- Nouveaux biomarqueurs pour un diagnostic précis et précoce
- Nouvelles thérapies innovantes et nouvelles orientations cliniques nationales et internationales
- Prévention et meilleure prise en charge de l'obésité
- Amélioration de la gestion des déficiences intellectuelles
- Surveillance renforcée grâce à des mesures cliniques et des biomarqueurs de fluides
- Meilleure compréhension de l'interaction entre l'obésité et les troubles cognitifs
- Visibilité et sensibilisation accrues à la trisomie 21 et à la recherche qui s'y rapporte

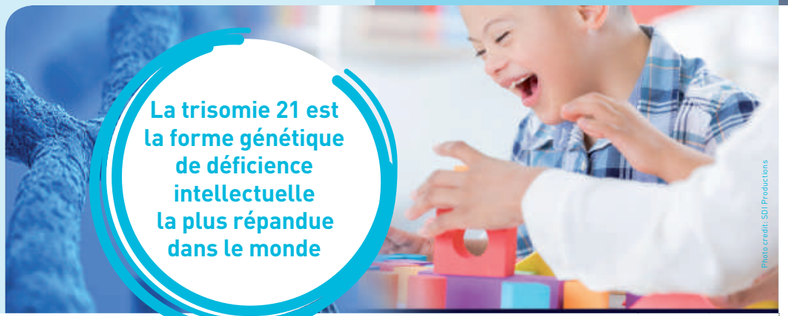

Photo credit: SDI Productions

La trisomie 21 est la forme génétique de déficience intellectuelle la plus répandue dans le monde

Photo credit: Izusek

NOTRE VISION

GO-DS21 vise à identifier et à valider de nouveaux mécanismes sous-jacents responsables de comorbidités observées dans la trisomie 21, principalement la déficience intellectuelle et l'obésité. Les personnes avec une trisomie 21 présentent souvent une combinaison de troubles mentaux (déficience intellectuelle, autisme ou troubles affectifs) et d'altérations physiques (anomalies cardiaques et hématologiques, obésité et anomalies gastro-intestinales).

À plus grande échelle, notre recherche permettra non seulement de mieux comprendre ces comorbidités dans la trisomie 21, mais aussi d'élucider le lien entre l'obésité et les troubles mentaux dans la population générale. L'obésité est un facteur de risque majeur pour de nombreuses maladies chroniques, contribuant à la mort de plus de 2,8 millions de personnes chaque année. De plus, l'obésité est souvent liée à un trouble cognitif, et augmente de façon spectaculaire dans les populations avec une déficience intellectuelle.

Ce projet de recherche avec une approche innovante et interdisciplinaire réunit des chercheurs fondamentaux et cliniques ainsi que des experts en physiopathologie, en bioinformatique et en intelligence artificielle. Go-DS21 s'efforce d'améliorer le diagnostic, le pronostic et le traitement précoces des affections qui coexistent dans la trisomie 21. Nous établirons de nouvelles recommandations cliniques et identifierons des interventions nouvelles et bien ciblées en début de vie pour prévenir ou au moins minimiser le développement de l'obésité dans le contexte de la déficience intellectuelle. Ces recommandations pourront également avoir des applications dans la population générale.

Annex 6: GO-DS21 project leaflet - ES

CONSORCIO DE INVESTIGACIÓN

12 instituciones y una organización de pacientes de 6 países con experiencia combinada en fenotipado de humanos y ratones, biología molecular y modelos informáticos

Organización de pacientes
Socios del consorcio
Coordinación científica

GO-DS21 EN POCAS PALABRAS

Título del proyecto:	**G**ene **O**verdosage and Comorbidities During the Early Lifetime in **D**own **S**yndrome
Fecha de inicio:	1 de enero de 2020
Duración:	60 meses (5 años)
Participantes:	12 instituciones de 6 países Europeos
Financiación CE:	6 millones €
Web del proyecto:	go-ds21.eu

CONTACTO

Coordinador científico:
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Gestión del proyecto:
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Síguenos en Twitter: twitter.com/GO_DS21

El presente proyecto ha recibido financiación del Programa de Investigación e Innovación Horizonte 2020 de la Unión Europea en virtud del Acuerdo de subvención nº 848077

Photo credits: DenKuvaiev, BlackJack3D

Dilucidando el vínculo entre discapacidad intelectual, obesidad y otras comorbilidades

¿POR QUÉ ES IMPORTANTE?

El síndrome de Down es la forma genética de discapacidad intelectual más común, con una incidencia de 1 de cada 1000 nacimientos y que afecta a más de 5 millones de personas en todo el mundo. Se caracteriza por una copia extra del cromosoma 21 humano (trisomía 21). Dentro de la población europea, la prevalencia del síndrome de Down sigue siendo alta debido al aumento de la edad materna y la falta de diagnóstico prenatal en determinadas poblaciones de riesgo. Además, el aumento de la esperanza de vida convierte a las personas con síndrome de Down en un segmento significativo de la población que necesita ser mejor investigada para mejorar su atención médica.

Condiciones mentales y físicas, como la discapacidad intelectual y la obesidad, aparecen en una tasa mucho más alta en personas con síndrome de Down en comparación con la población general. La obesidad en personas con síndrome de Down también aumenta el riesgo de desarrollar apnea obstructiva del sueño (un trastorno respiratorio potencialmente mortal), dislipidemia (una cantidad anormal de lípidos en la sangre), diabetes tipo-2 y problemas al andar.

Comprender las causas subyacentes y las vías biológicas de estas afecciones es por lo tanto de gran importancia para mejorar la salud de las personas con síndrome de Down. A pesar de la complejidad, la causa genética del síndrome de Down es clara: los genes del cromosoma 21 están presentes en exceso (3 copias en lugar de 2). Esto nos ayudará a aclarar los mecanismos subyacentes de la obesidad y otras comorbilidades.

IMPACTO EN EL MUNDO REAL

GO-DS21 utiliza datos nuevos preclínicos y clínicos procedentes de cohortes clínicas existentes además de generar nuevos datos. Los datos provienen de la llamada "red de comorbilidades del síndrome de Down", que incluye datos de cohortes en el Reino Unido (más de 6000 casos), Francia, España y otras tres clínicas europeas. Junto al conjunto de datos clínicos, GO-DS21 investiga hipótesis relevantes para el proyecto en modelos animales preclínicos para una mejor comprensión del síndrome de Down. Exploramos cómo factores intrínsecos (genéticos y vías) y extrínsecos (ambientales y epigenéticos) conducen a comorbilidades comunes. Los resultados del proyecto también pueden ser aplicables a personas que padecen otros tipos de discapacidad intelectual.

- → Nuevos biomarcadores para un diagnóstico precoz y preciso
- → Nuevas terapias innovadoras y novedosas recomendaciones clínicas a nivel nacional e internacional
- → Prevención y mejor manejo de la obesidad
- → Mejor manejo de la discapacidad intelectual
- → Monitorización mejorada a través de mediciones clínicas y biomarcadores en fluidos biológicos
- → Mejor comprensión de la interacción entre obesidad y deterioro cognitivo
- → Mayor visibilidad y conciencia sobre el síndrome de Down y la investigación relacionada

El síndrome de Down es la forma genética de discapacidad intelectual más común en el mundo

Photo credit: SDI Productions

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NUESTRA VISIÓN

Tenemos como objetivo identificar y validar nuevos mecanismos causales subyacentes a las enfermedades comórbidas, principalmente la discapacidad intelectual y la obesidad, en el síndrome de Down. Las personas con síndrome de Down suelen tener una combinación de trastornos mentales (discapacidad intelectual, autismo o trastornos afectivos) y deficiencias físicas (defectos cardíacos y hematológicos, obesidad y anomalías gastrointestinales).

Nuestra investigación no solo aumentará la comprensión de estas comorbilidades en personas con el síndrome de Down, sino que también aclarará el vínculo entre la obesidad y los trastornos mentales en la población general. La obesidad es un factor de riesgo importante para muchas enfermedades crónicas y contribuye a la muerte de >2,8 millones de personas/año. Además, la obesidad está relacionada con el deterioro cognitivo, aumentando drásticamente en las poblaciones con discapacidad intelectual.

El enfoque innovador de este proyecto interdisciplinar que une a investigadores básicos y clínicos, y a expertos en fisiopatología, bioinformática e inteligencia artificial. GO-DS21 pretende mejorar el diagnóstico temprano, el pronóstico y el tratamiento de las afecciones que coocurren en el síndrome de Down. Estableceremos nuevas recomendaciones clínicas e identificaremos intervenciones novedosas y focalizadas en las etapas tempranas de la vida para prevenir o al menos minimizar el desarrollo de la obesidad en el contexto de la discapacidad intelectual.