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EUROPEAN
DOWN SYNDROME
ASSOCIATION

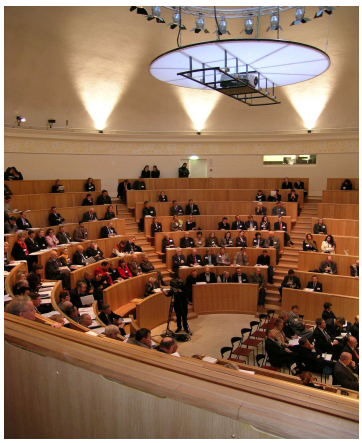
2ème Journée Mondiale de la Trisomie 21

TRISOMIE 21 EN MOUVEMENT

DOWN SYNDROME ON THE MOVE

23 - 24 MARS 2007

RESUMES / ABSTRACTS



COLLOQUE EUROPEEN

Grand Amphithéâtre
Muséum d'Histoire Naturelle

57, rue Cuvier
75 005 Paris, France

Sous le Haut Patronage de
Nicolas ABOUT, Président de la Commission
des Affaires Sociales du Sénat

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(anglais, français, espagnol)

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Dr. Renaud Touraine, Trisomie 21 France

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COMITE D'ORGANISATION

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L'Association pour la Recherche sur la Trisomie 21 a été créée en 1990 à l'initiative d'un groupe de chercheurs travaillant à l'hôpital Necker sur la trisomie 21. Elle est désormais principalement une association de parents qui est administrée par un conseil d'administration et un bureau émanant de ce conseil et dont les missions sont :

- Informer sur la Trisomie 21 grâce aux « Nouvelles du chromosome 21 »

14 numéros d'environ 10 pages, depuis 1995, traitant à la fois de données scientifiques et médicales sur la trisomie 21 et un petit livre publié en janvier 2005 qui rassemble les principaux articles publiés .

- Soutenir les programmes de recherche concernant les aspects fondamentaux, cliniques et thérapeutiques comme pour les autres maladies génétiq**ues**

Pour cela son conseil scientifique a permis à l'AFRT de subventionner quelque projets de recherche depuis 1998, sous forme de prix, bourses post-doctorantes ou doctorants et de subventions pour des projets de recherches spécifiques**.**

Nous disposons aujourd'hui d'une somme permettant à notre Conseil Scientifique de pouvoir continuer son travail pour susciter la recherche sur la Trisomie 21 en particulier sur les problèmes de sommeil et de phanères (peau et de cheveux) à la demande des parents de notre bureau.

- Participer à des colloques

L'AFRT a participé à de très nombreux colloques en France et en Europe sur la trisomie 21. L'AFRT est depuis 2005 membre de l'European Down Syndrome Association (EDSA) et de DSI (Down Syndrome International) dont le congrès aura lieu en juillet 2006 à Vancouver.

Elle a le 21 Mars 2005 tenu à la Mairie du 5 ème arrondissement à Paris la ***1ère Journée Nationale de la Recherche sur la Trisomie 21*** **« Du patient à la recherche, mieux comprendre pour mieux aider ».**
Depuis et à l'initiative de l'AFRT, la journée du **21 Mars** (3/21 ou 21/3 pour 3 chromosome 21) a été choisie par les instances internationales pour être désormais : **La Journée Mondiale de la Trisomie 21**

Nous avons tenu le **21 mars 2006** avec de nombreuses associations concernées par la trisomie à la Mairie du 5 ème arrondissement à Paris la ***1ere Journée mondiale de la Trisomie 21*** avec comme thème **« Comment appréhender et tenter de guérir le handicap mental « .** Participaient à cette journée Mr. P. Gohet, délégué interministériel aux personnes handicapées ainsi que Mr. PF Gachet, du Ministère de l'Education Nationale.

PROGRAMME

PROGRAM

Vendredi / *Friday* 23

10h30 **Bienvenue et discours d'ouverture** / *Welcome and opening addresses*

« 2007 : L'année européenne de l'égalité des chances »
"2007: The European Year of Equal Opportunities"
Modérateurs / *Chairpersons* : P. de Vismes et X. Jardin

La Convention internationale globale et intégrée pour la protection et la promotion des droits et de la dignité des personnes handicapées / *International globaland integrated for the protection and the promotion of rights and dignity for disabled persons*
J.L. Simon, Région Europe et Groupement Français de l'Organisation Mondiale des Personnes Handicapées (France)

Equal opportunities in everyday life / *L'égalité des chances dans la vie quotidienne*
J. Perera, Universidad de las Islas Baleares (Espagne, *Spain*)

The European Year of Equal Opportunity - A political perspective / *L'année de l'égalité des chances - Une perspective politique*
P. Clarke, Down Syndrome Ireland (Irlande, *Ireland*)

A life different but meaningful and productive / *Une vie différente et cependant valorisante et productive*
L. Vislan, ALDO-CET (Roumanie, *Romania*)

Opportunities for persons with Down Syndrome in the Czech Republic / *Opportunités pour les personnes atteintes de trisomie 21 en république Tchèque*
I. Moyano, Association of Parents and Friends of Children with Down Syndrome (Prague, République Tchèque, *Czeck Republic*)

12h30 Repas / *Lunch*
Espace Esclangon, Jussieu

Session 1	**Compétences du petit enfant et thérapeutiques précoces** *Child abilities and early therapeutics*

13h45 Modérateurs / *Chairpersons* : A. Kopecka et B. de Fréminville

Le suivi médical des personnes porteuses de trisomie 21 / *Medical survey for Down syndrome persons*
A. Rasore-Quartino, Hopital Galliera (Italie, *Italy*)

Hearing impairment in children and adolescents with Down syndrome / *Déficits auditifs chez les enfants et les adolescents atteints de trisomie 21*
J. Murphy, University of Dublin (Irlande, *Ireland*)

Structured phonological awareness intervention programme / *Programme pour l'amélioration des compétences langagières*
S. Macedo & **L. Cotrim**, APPT 21 (Portugal)

Understanding motor development of children with Down syndrome; Introduction of a theoretical framework / *Comprendre le développement moteur de l'enfant atteint de trisomie 21 ; Elaboration d'un modèle théorique*
P.E.M. Lauteslager, 's Heeren Loo Midden-Nederland (Ermelo) (Pays-Bas, *The Netherlands*)

Ocular torticollis in Down syndrome */ Torticolis et aspects ophtalmologiques dans la trisomie 21.* (Exposé en espagnol, *Spanish presentation*)
J.J. Puig Galy, Diagnóstico y Terapéutica Ocular (Espagne, *Spain*)

Latéralité (main, pied, oreilles et yeux) chez les personnes porteuses d'une trisomie 21 / *Laterality (hand, foot, ear and eye) in persons with trisomy 21*
M. Carlier, Université Aix Marseille 1 (France)

16h00 Pause café / *Coffee break*

Session 2	**Comment réussir l'insertion scolaire et les apprentissages à la vie autonome /** How to improve school integration and autonomous life

16h30 Modérateurs / *Chairpersons* : J.P. Champeaux et C. Baccichetti

Inclusive education and communication skills / *Education intégrée et compétences dans la communication*
E. Wilken, University of Hanover (Allemagne, *Germany*)

Capacités d'apprentissage des enfants et adolescents porteurs d'une trisomie 21 / *Learning abilities of children and teenagers with Down's syndrome*
S. Frenkel, Université de Picardie Jules Verne (France)

Teaching reading to develop both literacy and spoken language skills / *Apprentissage de la lecture pour le développement des langages écrit et parlé*
S. Buckley, The Down Syndrome Educational Trust (Royaume-Uni, *United Kingdom*)

Intervention Program designed to promote Speech, Language and Communication / *Programme d'intervention pour l'amélioration de la communication et des langages parlé et écrit*
F. Ferreira & **T. Condeço**, APPT 21 (Portugal)

School integration data for disabled children in Spain / *Données sur l'insertion scolaire d'enfants déficients en Espagne.* (Exposé en espagnol, *Spanish presentation*)
R. Borbonés, Fundació Catalana Síndrome de Down (Espagne, *Spain*)

La scolarisation des élèves porteurs de trisomie 21 : Quels parcours ? Quelles compensations à l'école ? / *School integration for Down syndrome pupils: Which school courses and wich benefits?*
J. Pennaneac'h, Trisomie 21 Loire (France)

Construction de la Personne porteuse de trisomie 21 : Les jalons de l'enfance / *Self construction of Down syndrome person: The groundsof childhood*
B. Céleste, Université Paris 10 (France)

19h00 Concert

Piano et chants / *Piano and classic songs*
Julien Rézac et Raphaëlle Wagnez
(Amphithéâtre 24, Jussieu)

Entrée libre / *Free entrance* **– Dons acceptés** */ Accepted donations*

Samedi / *Saturday* 24

Session 3	**L'âge adulte : Auteur de son projet de vie** *Adulthood: Author of his/her own life project*

08h30 Modérateurs / *Chairpersons* : A. Rasore-Quartino et J. Costils

Growth and Puberty in Down Syndrome / *Croissance, puberté et syndrome de Down*
J. Murphy, University of Dublin (Irlande, *Ireland*)

Building self – esteem / *Construction de l'estime de soi*
C. Baccichetti, Baccichetti foundation for Down syndrome (Italie, *Italy*)

The right to love. From a mother's diary / *Le droit d'aimer. Propos à partir du journal d'une maman*
R. Sneh, YATED (Israël, *Israel*)

The challenge of employment for young people with Down's syndrome / *Combat pour l'emploi des jeunes personnes atteintes de trisomie 21*
J. Perera, Universidad de las Islas Baleares (Espagne, *Spain*)

Présentation d'un dispositif d'insertion professionnelle en milieu ordinaire de travail pour des personnes porteuses d'une trisomie 21 / *Presentation of a professional integration frame for Down syndrome persons working in a normal working place*
V. Lombal, Trisomie 21 Gard (France)

Pause café / *Coffee break*

Pour une vie plus autonome / *Increasing autonomous life*
M.C. Haelewyck, Université de Mons-Hainaut (Belgique, *Belgium*)

Building up a social network through meaningful leisure activities / *Construction d'un réseau social à l'aide d'activités de loisir*
C. Halder, German Down Syndrome InfoCenter (Allemagne, *Germany*)

Independent Living Program: "I'm Going Home", a life project to build new opportunities in personal independence / *Programme pour une vie autonome : « Je vais à la maison », un projet de vie pour construire de nouvelles opportunités*
P. Ruf, Down Syndrome Catalan Foundation (Espagne, *Spain*)

Independent Living: the Italian experience / Vie autonome : L'expérience italienne
C. Leonori, Associazione Italiana Persone Down (Italie, *Italy*)

12h00 Repas / *Lunch*
Espace Esclangon, Jussieu

Session 4	**Le présent et le futur des thérapeutiques** *The present and the future of the therapeutics*

13h30 Modérateurs / *Chairpersons* : N. Créau, J. London et R. Touraine

Osteoporosis prevention in persons with Down syndrome / *Prévention de l'ostéoporose chez les personnes atteintes de trisomie 21.* (Exposé en espagnol, *Spanish presentation*)
P. Zubillaga, Uliazpi Foundation (Espagne, *Spain*)

Mortality and dementia in persons with Down's syndrome. The impact of Apolipoprotein E / *Mortalité et démence chez les personnes atteintes de trisomie 21. Le rôle de l'Apolipoprotéine E*
A. Coppus, Epidemiology & Biostatistics (Hollande, *Holland*)

How to face Dementia in DS Population. A Neurologist expert in Dementia point of view / *Que faire face à la démence dans la population atteinte de trisomie 21. Point de vue d'un neurologue.* (Exposé en espagnol, *Spanish presentation*)
I. Hernández, Institut Català de Neurociències Aplicades (Espagne, *Spain*)

One carbon metabolism, Immunology, and Growth hormone in Down syndrome / *Métabolisme des composés monocarbonés, immunologie et hormone de croissance dans la trisomie 21*
C. Romano, I.R.C.C.S. Associazione Oasi Maria Santissima (Italie, *Italy*)

Analyse de la paraoxonase-I, un marqueur de protection contre l'athéroscoclérose dans la trisomie 21 / *Analysis of Paraoxonase-1, a marker of protection against atherosclerosis in Down syndrome*
N. Janel, Université Paris Diderot (France)

Autoimmune disorders in persons with Down syndrome / *Anomalies autoimmunes chez les personnes atteintes de trisomie 21*
M. Sustrova, Slovak Medical University (Slovaquie, *Slovakia*)

Pause café / *Coffee breakF*

DYRK1A, a chromosome 21 gene involved in central nervous system development with potential phenotypic effect in Down syndrome / *DYRK1A, un gène du chromosome 21 impliqué dans le développement du système nerveux central et ayant un effet phénotypique potentiel dans le syndrome de Down*
M. Arbonés, Center for Genomic Regulation - CRG (Espagne, *Spain*)

DYRK1A, une cible thérapeutique ? / Dyrk1a, a therapeutic target?
J.M. Delabar, Université Paris Diderot (France)

Neurogenesis impairment and cell cycle alterations in fetal Down syndrome and Ts65Dn mice brain / *Anomalies de la neurogénèse et du cycle cellulaire dans le syndrome de Down et les souris Ts65Dn*
E. Ciani, University of Bologna (Italie, *Italy*)

Trisomie 21 et maladie d'Alzheimer / *Down syndrome and Alzheimer disease*
M.C. Potier, CNRS (France)

siRNA knockdown of the amyloid precursor protein (APP) in the brain in vivo / *Utilisation de la technique de siRNA pour éteindre « in vivo » dans le cerveau l'expression du gène APP (précurseur de la protéine amyloïde)*
K.L. Moya, Ecole Normale Supérieure (France)

La spectroscopie de résonance magnétique dans l'exploration métabolique des maladies du cerveau / *Magnetic resonance spectroscopy for metabolic exploration of brain disease*
P.J. Cozzone, Université de la Méditerranée (France)

18h00 Conclusion générale et clôture / *Final comments and perspectives*

18h30 Assemblée générale d'EDSA / *EDSA general assembly*

RÉSUMÉS DES PRÉSENTATIONS

ABSTRACTS OF THE PRESENTATIONS

La Convention internationale globale et intégrée pour la protection et la promotion des droits et de la dignité des personnes handicapées

Jean-Louis SIMON[1]
president.gfph@dpi-europe.org

Région Europe et Groupement Français de l'Organisation Mondiale des Personnes Handicapées
45, rue Riquet / A. 664, FR-75 019 Paris, France

Abstract:
Le débat sur les droits des personnes handicapées est ouvert depuis 1950 aux Nations Unies, et sous une forme ou une autre, fait partie de son programme d'action de façon ininterrompue depuis bientôt une soixantaine d'années.

Le changement d'approche conceptuelle introduit par le mouvement des personnes handicapées a permis aux Nations Unies de **passer** en quelques années **d'une approche principalement influencée par le modèle médical** et génératrice de réadaptation, **à une approche globale basée sur les Droits de l'Homme** et génératrice de transformations sociales.

L'ouverture prochaine à la ratification par les États de cette nouvelle Convention des Droits de la Personne est l'objet d'une cérémonie le 30 Mars 2007, et ouvre ainsi de nouvelles perspectives d'action en apportant à toutes celles et à tous ceux qui restent aujourd'hui dévalorisés du fait de leurs caractéristiques, une identité valorisante basée sur ce qui relie les humains entre eux : Leur égale valeur.

References:
- Convention des Nations Unis pour la protection et la promotion des droits des personnes handicapées
- L'accès aux droits, article de la revue « Reliance » N° 23, Mars 2007

Keywords: Droits de l'Homme, convention, Organisation des Nations Unies.

[1] President.

Equal opportunities in everyday life

Juan PERERA[2]
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CENTRO PRINCIPE DE ASTURIAS UNIVERSIDAD DE LAS ISLAS BALEARES
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Abstract:
In 2007, a year that has been declared the "European Year of Equal Opportunities for All" by the EU, the European Parliament and Council of the European Union aim to promote a series of shared measures so that countries understand the concept of diversity and encourage policies directed at non-discrimination.

One person can unite several characteristics that attract discrimination, thus experiencing it to an even greater degree. One example is the case of disabled women.

People think that the disabled are different from the rest of the population because we see their disability before we see the actual person. Presupposing that someone is incapable, singling them out and regarding them with pity curbs their development.

In everyday life, over-protection prevents development, reducing human beings to a state of alienated infancy synonymous with incapacity.

With the aid of good teachers and specialists, a person with a disability can come to accept their limitations and cope with life by making the most of themselves.

A person with a disability reaches adulthood bolstered up by all the affection, autonomy, understanding, stimuli and respect that they received and hindered by all the things they did not receive.

[2] Director.

The European Year of Equal Opportunity - A political perspective

P CLARKE[3]
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Down Syndrome Ireland
3 Woodgrange, Drogheda, Co Meath, Ireland

Abstract:
The talk will give a political over-view of the year of equal opportunity detailing the 3 main aims.
It will outline the manner in which the EU has decided to deal with the year and detail the six grounds of discrimination that are deemed to be of importance to the EU.
There are four key objectives or the 4 "R"s Rights – Representation – Recognition – Respect.
Details will be given of some of the European initiatives that are scheduled for 2007.
Give an analysis of Ireland's activities as it relates to the above.

Outline the European Disability Forums Campaign for a European Disability Directive
And its fight against discrimination

Keywords: EDF, discrimination, directive.

[3] President.

A life different but meaningful and productive

Lilian VISLAN[4]
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Abstract:
We bear in mind not only the abilities and needs of each person with Down syndrome but also that the professionals and services which the community presents to them must offer resources, skills, opportunities for development in the social and work fields by means of which these people can feel included within social, cultural and professional life with full rights. Therefore, people with Down syndrome should say: "we live a life different from that of many people, but a meaningful and productive, full and dignified life".

The quality of life of any person is measured fundamentally in the following dimensions: emotional well-being, interpersonal relationships, material welfare, personal development, physical well-being, self-determination, social inclusion and rights. And we base our own criteria on these dimensions when we talk about people with Down syndrome.

The 2007 European Year of Equal Opportunities for All is an initiative leading the way to a bolder strategy seeking to give momentum to the fight against discrimination in the EU, as the Commission explained in a document, published in June 2005, called '**Framework strategy for non-discrimination and equal opportunities for all**'. During the Year, all discrimination grounds have to be treated in a balanced way and the different ways in which women and men experience discrimination on the grounds of sex, racial or ethnic origin, religion or belief, disability, age or sexual orientation have to be considered as well.

References:
Barto, 1998
Robert L. Schalock
European Commission
Maria Vislan

Keywords: Down syndrome, life.

[4] Executive chairwoman.

Opportunities for persons with Down Syndrome in the Czech Republic

Isidro MOYANO[5]
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Association of Parents and Friends of Children with Down Syndrome
Gen. Janouška 1060, 190 14 Prague 14, Czech Republic

Abstract:
Children born with DS in the Czech Republic in the late 1990's and in the last few years have had significantly better future perspectives than those who had been born under the conditions of the former communist regime until 1989. Since that time there has been important progress not only in the way that general Czech society perceives health disability, but also in the legislative side in different important areas for social integration of persons with disability, such as in medical care, education, social affairs and culture. However there are still many limitations for people with disability and specifically with DS and many things to be improved and goals to be achieved.
Our Association of Parents and Friends of Children with DS was officially registered as a civic non-profit organization in Prague in 1996, but its activities had been started by a group of parents in 1993 under the supervision of the director of a Special Kindergarten - Special Education Center in Prague. Its members are not only families with their child with DS, but also specialists and other interested people from all over the Czech Republic. In other Czech regions 8 other organizations supporting people with DS have been established since 1990, and a few other organizations supporting integration of persons with disability.
We are pleased to present a new documentary film titled "*I'm looking for a job - I have DS*" (directed by Mrs. Olga Strusková), which has had its première in Prague just a couple of days ago on the occasion of the World Day of Down Syndrome. This film has been shot at the instance of our Association as the 3rd part of a series of documentary films about lives of people with DS, following two previous films: the first film *"One more chromosome"*, about integration of children with DS within families, and the second film "*At school together"*, about integration of children with DS in ordinary primary schools (both of them directed also by Mrs. Olga Strusková).
This film focuses on families' worries related to their teenagers with DS who had been integrated in different kinds of schools, about their problems with finding a job, about practical integration of teenagers and adults with DS in society and about the overall quality of their lives. By means of different individual cases it shows the present reality, which still has very limited opportunities for them. It shows opinions of specialists and also of potential employers.

References:
-Magazin PLUS 21 (published by our Association), No. 3 / December 2006, pages 7-8: article by film director Olga Strusková about her film.
-Presentations and publications of Association of Parents and Friends of Children with DS (Prague).
-Documentary film *"One more chromosome"* (2000, directed by Olga Strusková)
-Documentary film "*At school together*" (2002, directed by Olga Strusková).

Keywords: DS, Czech Republic, perspectives, adults, job, documentary film.

[5] Member of the Board.

Le suivi médical des personnes porteuses de trisomie 21

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Abstract:

Il existe depuis plusieurs années de nombreux programmes de suivi médical . Les modifications qui se vérifient constamment dans la société rendent nécessaires leurs mises à jour périodiques. C'est surtout chez l'adulte et la personne âgée que l'on a assisté aux modifications les plus importantes des conditions de vie et c'est pourquoi il faut prêter particulièrement attention sur ces tranches d'âge.

Le suivi médical commence par la période prénatale, qui est un moment capital pour l'acceptation ou le refus de l'enfant à naître. Les techniques de diagnostic prénatal sont à présent très diffusées ; le conseil génétique doit être disponible, mais doit offrir toutes les options possibles, avec une approche non directive.

Après la naissance, la communication du diagnostic à la famille est très délicate, pour l'acceptation de l'enfant dans le cadre de sa famille le moins difficile possible.

Les cardiopathies et d'autres malformations doivent être recherchées pour être corrigées précocement. De même, il faut rechercher des défauts visuels ou auditifs éventuels. Ensuite, on doit envisager des contrôles cliniques et neurologiques périodiques, des examens de laboratoire pour la prévention des pathologies thyroïdiennes et auto-immunes, de la maladie coeliaque et des leucémies. Le programme vaccinal doit être le plus complet possible. Les rééducations doivent être entreprises précocement.

Les capacités motrices, langagières, sociales et adaptatives doivent être contrôlées à chaque visite. Une aide psychologique doit être offerte aux parents et aux frères et soeurs. Il faut souligner l'importance pour la personne atteinte de trisomie 21 de l'estime d'elle même, pour un développement harmonieux et pour l'intégration sociale qui débute à l'école et qui se continue avec un emploi potentiel. Pour l'adolescent et le jeune adulte, une attention particulière doit être prêtée à la prévention de l'obésité et au développement psycho-affectif et sexuel. Contrôles cliniques et neurologiques doivent être poursuivis à l'âge adulte, pour la prévention et le traitement des troubles neuropsychiatriques qui peuvent subvenir tels que : troubles du comportement, dépression, convulsions et syndrome autistique.

Le vieillissement est le plus souvent normal, quoique plus précoce que chez la population non trisomique. Après l'âge de 50 ans une démence semblable à la maladie d'Alzheimer peut survenir pour environ la moitié des personnes atteintes de trisomie 21 chez qui la progression de la maladie est souvent rapide. Jusqu'à récemment, les traitements utilisés ont été décevants. A présent, l'utilisation de médicaments anticholinestérasiques semble avoir des effets positifs sur la progression de la maladie.

En conclusion, on doit être convaincu de l'utilité du suivi médical des personnes atteintes de trisomie 21, afin de leur donner la meilleure intégration possible dans la société et la vie heureuse qu'elles méritent.

[6] EDSA President.

Hearing impairment in children and adolescents with Down syndrome

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University of Dublin, Trinity College
The National Children's Hospital, Department of Paediatrics, AMNCH, Tallaght, Dublin 24, Ireland

Abstract:
Hearing impairment is one of the most prevalent disabilities in children and adolescents with Down syndrome and is challenging to identify and manage.1,2 There is an increased prevalence of conductive, sensorineural and mixed hearing loss in adolescents and adults, which is progressive in nature. Early identification and effective management of hearing loss is needed to prevent unnecessary secondary handicap that can lead to poor speech, learning difficulties and associated behavioural and emotional problems.3-5 Present hearing screening may be unreliable in these children as it is difficult for them to cooperate and respond at an appropriately early age because of associated intellectual disability. Distortion product Oto Acoustic emissions (DPOAEs) are produced by the inner ear when stimulated by two primary tones presented simultaneously. This quick, non-invasive, objective test requires no patient feedback and could be very beneficial to this group shortly after birth and throughout childhood and adolescents.
Aims: This study was undertaken to determine: 1) the prevalence of impaired hearing within age groups in the children and adolescents with Down syndrome in the Eastern Health Board Region; 2) the age when first hearing screening was carried out in this group; 3) to assess the outer ear by otoscopy and evaluate the function of middle ear by tympanometry; 4) to assess the feasibility of the use of DPOAE as an audiological tool to screen cochlea function in this population as this has not previously been performed in a large group with developmental disabilities.
Methods: 394 children and adolescents (217 boys and 177 girls) aged from 2 months to 19 years, with a mean age of 9.3 years participated in the study. Hearing screening was performed in clinical settings in non-sound proofed rooms at nine medical educational centres, one special school, the National Children's Hospital and home visits were performed.
Results: This study found 75% of children had hearing abnormalities. These included abnormal findings in the outer ear on otoscopy (64.4%), in the middle ear by tympanometry (58%) and in the inner ear by DPOAE (10.7%). Parents reported that children had their first hearing screening at the median age of 1.5 years (range 2 months to 14.5 years), and very poor compliance to wearing of hearing aids.
Conclusion: The use of this combined 3 tier screening for assessing hearing function is feasible, rapid and non-invasive which was acceptable and well tolerated by this group. Hearing screening in this manner could be performed from infancy and throughout life.

References:

1. **Cunningham C, McArthur, K**. Hearing loss and treatment in young Down's syndrome children. *Child: Care, Health and Development*, 1981, **7**: 357-374.
2. **Roizen N**. Hearing loss in children with Down Syndrome: A review. *Down Syndrome Quarterly*, 1997, Vol. 2 (4): 1-4.
3. **Evenhuis HM.** Dutch consensus on diagnosis and treatment of hearing impairment in children and adults with intellectual disability, 1992, Vol. 40: 5 October 451-456.
4. **Yoshinaga-Itano C**. Early intervention after universal neonatal hearing screening: impact on outcomes. *Ment Retard Dev Disabil Res Rev*, 2003, 9(4):252-66.
5. **Sigman M, Ruskin E, Arbeile S, et al.** Continuity and change in the social competence of children with autism, Down syndrome, and developmental delays. *Monogr Soc Res Child Dev*, 1999, 64(1):1-114

Keywords: Down syndrome, hearing impairment, otoscopy, tympanometry, Distortion Product Oto Acoustic Emissions.

[7] Research Nurse Specialist / Lecturer / Dr Joan Murphy RCN, MSc, Dip Stats, PhD.

Structured phonological awareness intervention programme

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Abstract:
Phonological working memory serves to store speech-based information temporarily and process it along with information that has been retrieved from long-term memory [1].
Research strongly points to a specific weakness in phonological working memory in people with Down Syndrome, which impacts negatively upon the development of language and reading skills.
This Intervention Programme was developed from several case studies with children and youths with Down syndrome under direct intervention, since the year 2000. This new-fangled intervention program correlates: letter sound, letter identification and syllable writing, with image support on the first stages.
The Structured Phonological Awareness Intervention Programme, is based on strategies that draw on memory and visual processing skills, it should be used from an early age, together with the "Reading Programme for the Training of Language Skills", as a complement to the process of acquisition of reading and writing skills.

Stages of this programme:

1. Development of attention span and listening skills.
2. Sound discrimination
3. Segmentation exercises (syllabic segmentation; Identification of words that are spoken in segments by the adult)
4. Letter identification and letter naming.
5. Sorting tasks (identify alliterations, rhymes, first syllables).
6. Audio discrimination of phonemes (reconstruction of words through phonemic discrimination and syllabic segmentation/discrimination)
7. Syllabic reading

This programme was designed to enable the learner:

a) To acquire reading and writing skills in a short time.
b) To improve intelligibility in verbal articulation
c) To improve concentration span, especially where joined attention is concerned
d) To become more proficient in work memory and listening skills
e) Fosters learning motivation
f) Enables children learn how to read and write independently in a playful manner.

References:

Conners, Frances, "Phonological working memory difficulty and related interventions". In Speech and Language Intervention in Down Syndrome, edited by Jean Rondal & Sue Buckley, 2003.

Keywords: Phonological awareness, writing, intelligibility, reading, down syndrome.

[8] Psychologist at APPT 21. [9] Psychologist

Understanding motor development of children with Down syndrome; Introduction of a theoretical framework

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Abstract:
Down syndrome children have a disorder-specific motor development profile. To understand their specific way of motor development the theoretical framework Disturbances in the regulation of postural control' has been constructed, based on accurate observations and related to scientific literature. This theoretical framework is a motor development model in which the attention is focused on the effect of motor disturbances on the development of functional motor behaviour in daily activities.

Literature shows all Down syndrome children have reduced postural tonus. Added to this there excist a lack of stabilization of joints and of balance reactions, a reduced propriocepsis and an increased mobility of joints. This leads in every phase of development to problems in acquiring and maintaining postures and to an inadequate development of balance.
However, the child wants to move, and will compensate his motor problems. Motor ability becomes very static, there is a lack of movement and of variety of movement. As a result the child develops specific motor behaviour with an insufficient level of functionality in play and care situations.

On the base of this framework a specific physiotherapy treatment has been developed including the motor measuring instrument 'Basic motor skills of children with Down's syndrome' (BMS) and the treatment method 'Physiotherapy for young children with Down's syndrome' (Lauteslager, 2004).

References:
Lauteslager, P.E.M. (2004). Children with Down's syndrome: motor development and intervention. Amersfoort: 's Heeren Loo Zorggroep (ISBN 90-73038-27-8, available in Dutch, English, Russian and Romanian).

Keywords: Down syndrome, physiotherapy, motor development, early intervention.

[9] Ph.D.

Ocular torticollis in Down syndrome

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Abstract:
PURPOSE: To ascertain the prevalence of torticollis in patients with Down syndrome.

METHODS: 760 patients with confirmed Down syndrome underwent complete ophthalmologic examination, which included refraction, ocular motility and head position.

RESULTS: Torticollis was present in 75 patients (9,86%). 80% of them were found to have an up or toward a shoulder head tilt. In 22 patients (29,33%) no ocular reason for the abnormal head position could be identified. The most common ocular etiologies were unilateral fourth nerve palsy and nistagmus.

CONCLUSIONS: Abnormal head positions are more common in patients with Down syndrome than in general population. Some of them seem to have no ocular etiology. Superior oblique muscle paresis has been the most common ocular reason for torticollis in our group of patients.

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Puig J, Galán A, Wert A, Santos E, Maciá C. Torticollis in Down Síndrome. Down Síndrome Research and Practice 2006; 9 (3): 83.

Keywords: Down syndrome, torticollis, strabismus, nistagmus.

[10] Ophthalmologist / Medical Doctor.

Laterality (hand, foot, ear and eye) in persons with trisomy 21

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Abstract:

Laterality (hand, foot, ear and eye) was observed in 62 persons with trisomy 21. Manual laterality was assessed with a fifteen-item task administered two times and with a card-reaching task. Two independent age groups were formed: 7 to 10 years old and 11 to 34 years old. The two comparison groups included 184 typically developing persons and 39 persons with Williams Beuren syndrome. We confirmed earlier data. Individuals with trisomy 21 were more frequently left - or mixed - handed than typically developing persons. Left right foot preference and cross hand - foot preference were more frequent in the group with trisomy 21 than in the typically developing group. Overall the laterality profiles were not the same in the two genetic groups: the greatest differences were observed between typically developing persons and persons with Trisomy 21. It is probable than the two groups with one genetic disease displayed different patterns because of the specific feature of their genetic disorders.

References:

Carlier, M., Doyen, A.-L., Lamard C. (2006). Midline crossing: developmental trend from 3 to 10 years of age in a preferential card-reaching task. *Brain and Cognition*, 61, 255-261.

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Gérard-Desplanches, A, Deruelle, C., Stefanini, S., Ayoun C., Volterra, V., Vicari, S., Fisch, G., and Carlier, M. (2006). Laterality in Persons with Intellectual Disability. II. Hand, foot, ear and eye lateralities in children or adolescent and adults persons with Trisomy 21 and Williams Beuren syndrome. *Developmental Psychobiology*, 48, 482-491.

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Inclusive education and communication skills

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Abstract:
Inclusive Education will not separate and then integrate the child but on the contrary help him to participate in all areas of normal daily life. One of the main aspect for inclusion is the abilities of the child to communicate. If we not only look at the stars with Down`s syndrom but at the average developing child and at the very slow learners, it is necessary to offer alternative and augmentative communication as early as possible. Thus the child will be able to ask questions, to say what he wants and to share in decisions – skills, which are important at home, in kindergarten and school.
Sign-language is especially for the young child a key to understanding the world – and for his parents and peers it is a window to the world of the child. A relevant additional aspect is that during the time when the biological window for learning words and grammar is open, the child has an alternative instrument – and when he is able to speak he has only to switch the mode.
In school sign-language and additional elektonic augmentative and alternative communication aids with natural or synthetic speech can support reading skills. They are also an important help for the child who is not able to speak or to speak understandably in order to participate in discussions and other classroom-activities.

[12] Professor.

Learning abilities of children and teenagers with Down's syndrome

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Abstract:
Progress in biomedical research confirm the fact that actually it is unfortunately not possible to use a genetic treatment in order to remove (or even to attenuate) the harmfull effects of this chromosomal anomaly (trisomy 21) on the intellectual functionning of people who are carrying it. However, it is possible to act on the conative and environmental determinants; in particular usying a program of cognitive remediation. The main goal of these type of program is to develop the pupil's intrinsic motivation as to develop and generalizing uses of the executive functions.

We propose to expose and make an appraisal of the nature as well as the effects of our training in rehearsal strategy. Its principal goal was the spontaneous use of a mnemonic strategy: active rehearsal (*i.e.*, cumulative repetition of the verbal items to memorize).

Fifty two children and teenagers with Down's syndrome took part in a six-weeks training. Effects are assessed immediately (post-test 1), 3 weeks (post-test 2) and 7 weeks (post-test 3) after the last training session. Results are exposed concerning 42 participants.

Keywords: Down's syndrome, rehearsal, memory, training.

[13] Doctor in Developmental psychology, Ph.D. / Psychologist.

Teaching reading to develop both literacy and spoken language skills

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Abstract:
This paper will present a review of the current literature on reading development in children with Down syndrome in both preschool and school years. It will highlight the evidence that reading ability is often a relative strength in relation to other cognitive, academic and language skills and explore the ways in which reading skills develop for this group of children. The presentation will include video of early readers. Drawing on the literature to date, and work in progress, recommendations will be made for developing effective reading instruction in preschool and school years, which will capitalise on the visual learning strengths of the children and maximise the benefits for their spoken language.

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Groen, M. A, Laws, G., Nation, K., & Bishop, D.V.M., (2006) A case of exceptional reading accuracy in a child with Down syndrome: Underlying skills and the relation to reading comprehension. *Cognitive Neuropsychology*, 2006, 23 (8), 1190–1214.

Hulme, C (2005) Reading development in children with Down syndrome: relationships with oral language and phonological skills. Paper presented at 4th International Confernce on Developmental Issues in Down Syndrome. University of Portsmouth.

Buckley, S. J., (2005) Teaching reading to teach talking. Paper presented at 4th International Confernce on Developmental Issues in Down Syndrome. University of Portsmouth.

Keywords: Reading, phonological skills, speech, language, reading instruction.

[14] Director for research.

Intervention Program designed to promote Speech, Language and Communication

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Abstract:
The APPT21 has developed, since 1992, an Intervention Program aimed at promoting communication in children with Down Syndrome. The Programs methodology has been improved over the years with new strategies, intended to develop cognitive and communication skills based upon their developmental characteristics.
In our presentation, we propose to highlight the main features of the two part Program, its methodology and strategies and the software developed to facilitate children performance.

1) The "PreComunication" Program was designed to promote the early communication skills of children with T21. This Program includes information about: what is Total Communication; the early use of natural gesture and enhancement of joint attention. This first Program also presents "Sign Language Workshop".
The "Sign Language Workshop" is an interactive multimedia application intended to:
- Be an essential support for parents, educators and therapists working with children with Down's syndrome in early intervention programs,
- Assign, for each sign, a set of information in diverse formats (text, sound, image, video), making possible personalized research.

2) The 2nd part "Learn To Read to Support Language" was designed to enhance speech, language and early reading skills. This Program includes information about: the methodology and the skills checklist, a vocabulary checklist to assess and monitor children's comprehensive and expressive language; how to promote phonological awareness. The 2nd part of the Program also introduces "Mimocas Games".
"Mimocas Games" is an interactive educational software game, especially designed for use with children with Down syndrome. Its key objectives are:

a) To promote comprehensive and expressive language development and verbal short-term memory;
b) To accelerate mastery of grammar and sentence building;
c) To teach youngsters with Down's syndrome how to read by means of a visual learning process.

Keywords: Communication, Down Syndrome, language, multimedia applications, programs.

[15] MGeog; Volunteer worker at APPT21 since its foundation / Responsible for APPT21 website / Co-responsible for the development of all multimedia applications developed by APPT21.

[16] Psychologist.

Algunos datos entorno a la inclusión del alumnado con discapacidad en el Estado Español

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Abstract:
La inclusión escolar del alumnado con discapacidad en el territorio del Estado Español es una realidad que se está llevando a cabo con éxito a pesar de las dificultades que han ido surgiendo en el proceso de su implantación.
La exposición que se va a presentar en el Meeting European de la European Down syndrome Association (Edsa) va a tratar de diversos aspectos que se citan a continuación.
En primer lugar, se expone cuál es el marco legal y normativo común a todo el Estado Español ; se citan las leyes generales que han hecho posible la inclusión escolar de los alumnos con discapacidad:
LISMI (1982) Ley de Integración social de los minusválidos,
LOGSE (1990) Ley de Ordenación general del sistema educativo y
LOE (2006) Ley de Ordenación Educativa.

A continuación, se van a presentar una serie de datos entorno a la escolarización del alumnado con necesidades educativas especiales (NEE) según sea el centro al que asisten (ordinario o especial) en las diferentes Comunidades Autónomas. En el Estado Español las competencías en materia educativa están transferidas a las Comunidades Autónomas lo que conlleva que existan diferencias notables entre unas y otras.
Finalmente, se hablará de los Servicios Educativos existentes en Catalunya, como órganos de apoyo a los centros docentes en materia de atención a la diversidad y de los aspectos más relevantes del proceso de inclusión de alumnado con síndrome de Down (SD).

References:
-J. M. JARQUE (2007) no publicado.Gráfica de escolarización del alumnado con discapacidad. Resumen cursos escolares del 1981/82 hasta 2005/06.
-Ministerio de Educación y Ciencia. Estadística de la Enseñanza en España niveles no universitarios. Oficina de Estadística. Gráfica del alumnado con NEE en integración y en centros específicos. Curso 2003-04.
- Departament de Educació. Generalitat de Catalunya. Servei d'Estadística i Documentació. Estadística de l'ensenyament. Curs 2004-2005.

Keywords: Marco legal y normativo, inclusión educativa, alumnado con SD, escolarización en centros ordinarios, servicios educativos.

La scolarisation des élèves porteurs de trisomie 21 : Quels parcours ? Quelles compensations à l'école ?

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Abstract:
Le système scolaire français et le secteur du handicap se sont construits sur le mode de la séparation. Les références culturelles et sociales des professionnels se sont donc élaborées en cohérence avec cette histoire. La demande de scolarisation des élèves en situation de handicap est avant tout une demande familiale qui vient peser sur les acteurs traditionnels de l'école et du champ du handicap. Les modalités de scolarisation sont donc traversées par la nécessité de donner un sens à la présence d'élèves perçus comme trop différents du groupe classe. La définition des accompagnements et des modes de compensation s'en trouve donc aussi affectée.

La loi du 11 février 2005 propose que l'on s'attache maintenant au « comment faire » plutôt qu'au « pourquoi faire », or il me semble que le travail d'explicitation du fondement de la scolarisation en milieu ordinaire de toute personne en situation de handicap et en particulier des personnes porteuses de trisomie 21 reste encore largement à faire. La définition des moyens de compensation ne pourra être pertinente que lorsque l'ensemble des acteurs aura une perception plus claire du sens et des enjeux portés par cette question.

[17] Directeur.

Construction de la Personne porteuse de trisomie 21 : Les jalons de l'enfance

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Abstract:
Dans le cadre théorique Wallonien, la « Personne » au sens psychologique émerge de l'adolescence en se positionnant en tant que telle par la propre définition de ses orientations, de ses choix et de ses valeurs. Il ne s'agit alors que de l'aboutissement d'un processus de construction qui s'enracine dans la toute petite enfance et s'ancre dans les relations construites avec autrui.
En nous appuyant sur 2 exemples :

- L'interprétation des réflexes gusto-faciaux,
- La construction de l'identité sexuée,

nous montrerons les entraves mises à cette construction dès les premiers jalons, pour les individus porteurs de trisomie. Les résultats de ces recherches témoignent de la difficulté de tous les « autrui » (familiaux et non familiaux) à donner au jeune enfant porteur de trisomie les points de repères nécessaires à l'émergence de sa « personne ».
En conclusion, nous évoquerons quelques pratiques utilisées dans l'accompagnement psychologique des familles et des personnes trisomiques pour pallier cette difficulté.

References:
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Céleste B., (1997), L'autonomie, une demande paradoxale, *trisomie 21, le défi de l'autonomie*, actes des journées nationales de la Fait21, Clermont-Ferrand,

Céleste, B. (1999), La spécificité du bébé trisomique dans ses manifestations émotionnelles, *Nouvelles du chromosome 21 A.F.R.T.*, 6, mai.

Céleste, B(2006), Garçon ou fille ? une construction difficile pour l'enfant trisomique. In *Enfant en développement, famille et handicaps*, érès, Ramonville Saint-Agne, 139-150.

Keywords: Trisomie 21, identité sexuelle, autonomie.

[18] Vice-Président professionnel Trisomie21-France / Vice-président Université Paris10 / MC en Psychologie du développement.

Growth and Puberty in Down Syndrome

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Abstract:
Short stature is well recognised in Down syndrome (DS). The aetiology of this growth pattern with a reduced pubertal growth spurt is not fully understood.1,2 Associated conditions, such as hypothyroidism, sleep related upper airway obstruction or coeliac disease may contribute to poor growth and must be excluded promptly in order to optimise the already compromised growth potential.3-5 Those with DS have an increased incidence of overweight and obesity, which may be associated with significant adverse health outcomes. The timing of puberty varies in this population, requiring anticipation and education.
OBJECTIVES: To assess growth and pubertal development in children and adolescents with DS in Ireland; compare with the general population and similar groups internationally; and also determine the effect of cardiac disease on growth.
METHODS & MATERIELS: A cross-sectional study of 394 children with DS was undertaken in Ireland. Height, weight and head circumference measurements were performed and clinical assessments of pubertal development were recorded.
RESULTS: Height and head circumference were 2 standard deviations (SD) below the mean of the general population but median weight was similar. Body mass index was greater than the 97th percentile in 31% of those over 10 years of age. Mean age of menarche 12.6 years (range 9-15 years) was earlier than the general population. Associated cardiac disease showed no effect on height.
CONCLUSIONS: Children with DS are shorter, have a smaller head circumference but similar weight as the general Irish population indicating a significantly greater body mass index (BMI). Monitoring growth requires DS specific growth charts and targeted weight management programmes. Puberty requires early anticipation, child and parental education and family support.

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4. **Stebbens VA, Dennis J, Samuels MP, et al**. Sleep related upper airway obstruction in a cohort with Down's syndrome. *Arch Dis Child*, 1991, 66: 1333-1338.
5. **George EK, Mearin ML, Bouquet J, von Blomberg BME, et al**. High frequency of celiac disease in Down syndrome. *J. Pediatr*, 1996,128: 555-57.

Keywords: Down syndrome, growth, overweight, obesity, puberty.

[19] Research Nurse Specialist / Lecturer / Dr Joan Murphy RCN, MSc, Dip Stats, PhD.

Building self -esteem

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Abstract:
It is important that young persons with Down syndrome feel confident about decisions they make and about the abilities they possess. In order to promote self confidence is important

1) Do not view individuals with Down syndrome as helpless. Ross (1964) reports that "the child derives bis self-image from the attitude those around him hold and manifest toward him. Because of this process of identification, if the child is seen as helpless, he or she will internalize that view. Barsch (1961) suggests that even the siblings of a child with mental retardation will imitate their parents' attitudes toward that child. It is imperative then, that children with Down syndrome grow up in an environment where they are viewed as able and worthwhile. Expressions of support and confidence in a child's abilities will strengthen the early development of healthy self-esteem.

2) Allow independence, autonomy, and self-motivation to grow.

3) Help persons with Down syndrome to experience success.

The right to love: From a mother's diary

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Abstract:

In using the term "The Right to Love" my intention is to the right to develop a relationship with another person, especially an intimate relationship with someone from the opposite sex.

This basic right belongs to everyone, but is not possible for most individuals with intellectual disabilities to realize. The major reason for this is the reality that members of this group need guidance and support in developing social relationships and in realizing a sexual relationship.

A small minority of individuals with mild retardation are sucessful in developing relationships on their own. Most individuals functioning at the moderate level of retardation, and below, suffer from feelings of isolation, even if they have difficulty expressing these feelings.

As far as I know a study of social and sexual relationships of people with mental retardation, living in the community, has not been undertaken.

My conclusions are based on my close relationship of many years with my son who is now 33 years old and has Down syndrome. Sexual needs remain unsatisfied since the individuals need guidance and there are very few qualified sex therapists, I estimate less than 10 in the country, who are trained to treat individuals with intellectual disabilities.

Many of these individuals, who become agitated because of the lack of sexual release, are prescribed psychiatric medications to calm them down and thus they are "punished" twice. On the one hand they do not achieve any sexual satisfaction and on the other they are prescribed unnecessary medication.

When my son was young I was guided by professionnals to focus on developing his intellectual abilities. This was a mistake, about which I would like to warm both parents and professionals. It is necessary to begin from an early age to teach the children to develop friendships and to support their sexual development. Those responsible for the care and treatment of individuals with mental retardation do not deny the "right to love" or to intimacy, but they do not do enough to help realize this right.

In Jerusalem a club designed to help educate young people with social needs about relationships and intimacy has recently opened and I will present details about this activity in this session.

[20] Vice Présidente.

The challenge of employment for young people with Down's syndrome

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Abstract:
The best thing we can do today for a young person with Down's syndrome is to guide them toward employment, teach them to work, find a job for them, and offer support in their work while they need it.

Given the diversity that characterizes Down's syndrome, we base this task on a model that is giving very good results.

When the students finish primary education at an ordinary school, they have to choose one of two options: to go on to secondary education or to study equivalent courses or vocational training, known in Spain as "social guarantee" schemes or the Transition to Adult Life.

Those who are unable to pass these courses are directed toward Occupational Centres (similar to the sheltered workshops of the French system). Others with a higher capacity are directed toward Special Employment Centres, once they have undergone corresponding screening procedures, while the most capable, well-prepared ones are encouraged to work in ordinary businesses under the "supported employment" system.

What marks this model out is the fact that we consider all these levels to be WORK and all the young people engaged in them to be WORKERS, as if the different levels were steps up a LADDER. They are open-ended, temporary, permeable levels. Workers can go up or down a level depending on their capacity, interest, training, performance, motivation or the support they receive. All these levels are remunerated in some way.

The end goal is for workers to be at the highest possible level at all times, depending on their capabilities, motivation, adaptation and performance.

[21] Director.

Présentation d'un dispositif d'insertion professionnelle en milieu ordinaire de travail pour des personnes porteuses d'une trisomie 21

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Abstract:
Un dispositif d'insertion spécifique

Le dispositif d'insertion que nous présentons est envisagé comme **un processus dynamique et évolutif,** fondé sur la notion **d'employabilité et de capacités.**

Comment faire en sorte que la personne porteuse d'une trisomie soit reconnue « employable » par l'entreprise , c'est à dire compétente et capable de travailler dans la durée

Et

Comment faire en sorte que l'entreprise du milieu ordinaire soit « capable » d'intégrer une personne porteuse de trisomie, qu'elle puisse être considérée comme un lieu possible d'emploi par la personne trisomique et comme un lieu où elle puisse s'envisager dans son devenir et son évolution

Plusieurs dimensions seront prises en compte:
- celle liées au développement de compétences des deux acteurs (personne handicapée/ entreprise): compétences techniques, professionnelles et comportementales,
- celle relative aux conditions environnementales (personnel et professionnel),
- enfin celle relative à l'organisation du travail et au partage des compétences.

Keywords: Trisomie 21, travail, formation, insertion professionnelle, accompagnement.

[22] Chargée de mission.

Pour une vie plus autonome

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Abstract:
Les concepts de normalisation et d'intégration sont aujourd'hui assortis des termes de participation, d'implication et d'influence, termes derrière lesquels se dessine une exigence croissante d'écoute de l'usager.
La discussion porte désormais sur la manière de mettre en place les conditions d'un dialogue entre toutes les personnes concernées (usagers, parents, personnel, collectivité locale). Ce dialogue est d'autant plus important que les usagers, en tant que groupe, sont beaucoup plus visibles qu'avant, donc plus présents dans la conscience de la population.
Nous aborderons les dimensions relatives à la qualité de la participation des bénéficiaires. La singularité des échanges passe par une bonne connaissance de soi, de ses forces et de ses faiblesses ainsi que de son histoire personnelle insérée dans un réseau de relations. Il s'agit de pouvoir aborder ce que l'on perçoit, ressent et fait.
La parole authentique et personnelle peut être favorisée par les contextes de vie mais elle nécessite également des méthodologies adaptées pour faciliter l'expression des personnes présentant un retard mental : trajectoire de vie (Gosset et al., 2000) ainsi qu'une meilleure connaissance d'elles - même et de la gestion de leur propre vie : l'autodétermination (Bara et Haelewyck, 2004) la prise de décisions (Hanot, 2002), l'autorégulation (Haelewyck et Nader-Grosbois, 2005) qui peuvent mener à des apprentissages ciblés. Ceux-ci peuvent être entrepris tout au long du cycle de vie et ce, y compris à l'âge adulte.
Un modèle axé sur la participation des usagers ne signifie pas que ce dernier a simplement le droit de s'exprimer et d'influer sur le cours de son existence, mais également qu'en tant que citoyen il doit bénéficier de la possibilité de participer à part entière à la mise en forme de la société dans laquelle nous vivons tous. Adopter cette démarche c'est permettre à l'usager de pouvoir choisir sa vie et l'accompagner en ce sens au sein de nos réunions de travail.

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Keywords: Autodétermination, autorégulation, identité personnelle.

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Building up a social network through meaningful leisure activities

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Abstract:
In the lifeplanning of people with Down syndrome the focus has been mainly on school education and preparation for work. The area of leisure has been neglected. However, an adult with Down syndrome – usually not fully employed – has many hours a day which he or she could use for activities other than work.
In recent years we have had to deal with an increase in problems of adults with Down syndrome, such as withdrawal, losing interest, decrease of skills, lethargic behaviour, depressive mood etc.
We found that many of these people lead very dull, uneventful and lonely lives. They have few personal contacts – mostly only with family and some paid helpers - no friends, no acquaintances.
They have few opportunities to go somewhere and take part in activities in their communities. They have a lot of free time, but show more and more passive behaviour.

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Independent Living Program: "I'm Going Home" a life project to build new opportunities in personal independence.

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Abstract:
The Independent Living Program *"I'm Going Home"* was created by the Down Syndrome Catalan Foundation in the year 2000 as an experimental social welfare program in Catalonia. One year after we began work on this project, the government gave its full support by passing a new law guaranteeing economic resources for models of attention such as ours. Now after the seven years that our program exists, those persons with intellectual disability or Down's syndrome have an autonomous life in their own homes. With their rights ensured, we see that change *is* possible to social policies.
Our service offers to persons with intellectual disability the opportunity to create their own lifestyle, designing their own individual support and planning for their own increased self-management. Our goal is to help satisfy the individual needs and requirements of every person in their own environment.
For these past seven years we have seen the creation of many different models of services and programs giving support to independent living all over the world. This important change inspired by the Independent Living Movement or MVI represents and reclaims for all persons the right of self-determination. People with intellectual disability have had minor representation in this broad movement because their voice, demands and expectations are principally represented by others: family, professionals or organizations. In fact, many times we see most of these models as just sophisticated planning of existing educational programs, suspiciously far from the real wishes of the individual.
With the international community supporting equal opportunity and quality of life for all persons, we must ask ourselves what our contribution can be and how our own organizations can promote this equality.

Convention on the Protection and Promotion of the Rights and Dignity of Persons with Disabilities. United Nations. New York, 2006.
Article 19: Living independently and being included in the community

(a) Persons with disabilities have the opportunity to choose their place of residence and where and with whom they live on an equal basis with others and are not obliged to live in a particular living arrangement;

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Vidal Garcia Alonso, J (Coord.): "El movimiento de vida independiente. Experiencias internacionales." Fundación Luis Vives. Madrid, 2003.

Keywords: Independent living, self-determination, MVI, quality of like.

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Independent Living: the Italian experience

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Abstract:

The all process of growing up and self-development can be seen as a gradual transition from being dependent to being independent. The all process can be said to be complete when the child becomes an adult and citizen in all his aspects, able to express his rights and able to work and have equal relationships.

But a child with special needs encounters two types of problems that make it difficult for them to become autonomous. Not only is there the difficulty represented by the handicap, but there is also the ambivalent attitude of society. Very often people in contact with children with special needs, develop an over-protective attitude which hinders the child's acquisition of independence.

It almost seems as if people want to compensate the unease created by the deficit by means of heightened affection and more permissive attitudes, or that, because of the deficit, the child is held to be generally incapable and thus in need of help and of someone who stands in for him on every occasion. In spite of these difficulties they can and do achieve autonomy.

An idea has gained ground in recent years, among workers who deal with the mentally retarded - namely the conviction, espoused with increasing strength, of the importance of educating these subjects to be autonomous as part of their personal development and their integration into society.

It is now proved that a good level of independence among people mentally retarded is a necessary requirement for this integration, sometimes it is difficult to reach the aimed goal, especially within the family unit, because teen-agers, either with or without DS start to show a need of detachment from their families and they start to be intolerant towards their requests.

On the other hand parents find awkward to accept that their "kids" are growing up this is when the education to the independence become particularly epoch-making.

With this in mind the AIPD has developed a special educational course with the aim of increasing autonomy in people with DS. The first course was organised with the Rome branch of the AIPD in 1989 and was designed for boys and girls ranging in age from 15 to 20. Since 1989 it has been repeated every year with an ever-greater participation and replicated in many other cities.

I felt it was necessary to mention this experience because through it has been possible to design a path, which has seen the participation and involvement of people with DS, educators and families towards the recognition of growing-up and self-esteem of themselves.

To the weekly meeting we added up week-ends of small groups and the first summer holiday which, once more in order to develop their independence, gave them the opportunity of learning new skills such as going shopping, running the house including cooking, and also organizing their free-time on their own.

Such a great experience gave people with DS the chance to be independent and responsible. their parents could try out the feeling of separation from their children.

It is essential now to emphasize the fact that we haven'y made any selection among users even if, as we all know, there is a large variability among people with DS.

All the families who had agreed with such an initiative were allowed to enrol their child and for each individual a "tailor-made" path towards autonomy has been designed. This has meant, at the end of the course, that someone was able to move around Rome without any problem; someone else became capable of walking little distances on his/her own orentering the main entrance of our "Everyone has got his/her own individual autonomy". This has been and still is the maxim that characterises the job of educators, with the faith that everyone would have been able to make a little step forward or, at least, to be able to maintain in the future what he/she had achieved.

As children grow up and after many debates with their parents, we started considering the whish and sometimes the need to live away from home..

First step was to think that it was necessary to consider the "going out" of the family towards a home group (or similar solution not just as a necessary event (due to the death of the parent or to impossibility to go on with the rest of the family) but as an autonomous choice of independent living.).

Osteoporosis prevention in persons with Down syndrome

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Abstract:
The prevention of osteoporosis is based on a number of hygienic, dietetic and pharmacological measurements, which share the objective of achieving an optimum level of well-mineralised bone mass during the period of development and its maintenance at safe levels during the rest of life. The final objective is to avoid the problems, mainly fractures, that are directly related to low levels of mineralization. There are publications that show that adult people with Down's Syndrome manifest levels of bone mineralization lower than what is considered normal in the general population.
The development of bones starts in the prenatal period, reaches its peak at the end of the second decade in life, and starts to decline from the age of 50 in women and 65 in men. In the development of the bones there are at least three significant factors: certain hereditary factors (that are not well known), physical factors related to muscular exercise, and dietetic factors. Amongst the latter a suitable energy intake with sufficient levels of proteins, calcium and vitamin D is essential. The typical diet in western cultures guarantees a sufficient contribution of energy, proteins and calcium. Alternative diets must be assessed in each case to guarantee that they contribute these nutrients in suitable proportions. In some population groups, amongst which is the Down population, there have been described insufficient levels of vitamin D.
Celiac disease is frequent in the general population and even more frequent in the Down's population. If not treated this is an important source of failure in the mineralization of the bone. This must be discounted in all cases by the determination of serological and genetic markers.
The data known so far, relating always to the general population, cannot be considered sufficient to catalogue the Down's population as special risk regarding osteoporosis. It is necessary to establish data on the Downs' population , particularly whether these data correspond with a greater frequency or gravity of fractures and problems related to osteoporosis.

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Keywords: Down syndrome, osteoporosis, vitamin D deficiency.

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Mortality and dementia in persons with Down's syndrome
The impact of Apolipoprotein E

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Abstract:
Aim: To study the effects of APOE on mortality and dementia in a longitudinal prospective study of a large population based sample of persons with Down's syndrome (n=425), demented and non-demented, aged 45 years and above (1).

Methods: A standardized assessment of cognitive, functional and physical status was repeated annually. If deterioration occurred, the participants were examined and the differential diagnosis of dementia was made according to guidelines produced by the Ageing Special Interest Group of the International Association for the Scientific Study of Intellectual Disabilities (IASSID)(2).
APOE genotypes were determined using Taqman allelic discrimination technology.

Results: When combining our cross-sectional data with those of other studies in a meta-analysis, we found APOE4 was significantly associated to the prevalence of dementia. The prospective data showed an increase in incidence of dementia in APOE4 carriers (one or two E4 alleles present). The frequency of APOE4 carriers decreased significantly after the age of 60 years, suggesting an increased mortality before this age. It is interesting to see which factors predict the survival in this study during the follow up.

Conclusion: Since dementia is the major cause of mortality in persons with Down's syndrome aged 45 years and older, an effect of APOE4 on dementia is in line with the reduction of APOE4 allele frequencies after the age of 60 years.

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2: Aylward EH, Burt DB, Thorpe LU, Lai F, and Dalton A. Diagnosis of dementia in individuals with intellectual disability. J Intellect Disabil Res 1997; 41 (Pt 2): 152-164.

Keywords: Down's syndrome, Alzheimer's disease, apolipoprotein E, co-morbidity, mortality.

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How to face Dementia in DS Population. A Neurologist expert in Dementia point of view

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Abstract:
Study background: New pathologies, such as dementia are appearing secondary to longevity in people with DS. 'Older' DS subjects have a higher risk of developing Progresive Dementia Syndrome (PDS). Alzheimer Disease (AD) and PDS/DS share the same neuropathlogic features and neurotransmitter disorders, and are linked to chromosome 21/APP. The treatment of cognitive decline and PDS in 'older' adults with DS has yet to be fully studied. Several studies have shown that cholinesterase inhibitors (ChEI) have beneficial effects in PDS/DS.
Objectives: Main objective: To evaluate the clinical benefits of specific Alzheimer's treatment, inhibitor acetilcholinesterase, Donepezil, dose range and tolerability in persons with PDS/DS, older than 40 years.
Secondary objective: To use the Severe Impairment Battery (SIB) and the Mini Mental State Examination (MMSE) as standard neuropsychological tests for dementia feasible to assess cognition in adults with DS
Material and Methods: 209 patients from "Fundació Catalana Dindrome de Down" database ≥ 40 years. 99 subjects were interviewed using Early Signs of Dementia Checklist (ESDC) by carers and referred to Fundació ACE. 45 subjects had ≥10 items affected on ESDC and could enter the study.
Primary efficacy was measured using the Dementia Questionnaire for Mentally Retarded Persons (DMR)
Secondary efficacy was measured by the Adaptative Behavior Scale Residential and Community (ABS-RC:2). Independly we use others NPS tools, MME and SIB. validate in Alzheimer's population and standarized in a clinical trials in AD.
Conclusions: New neurocognitive and functional tools should be designed and adapted in order to achieve early diagnosis of PDS in 'older' DS subjects. Both, MMSE and SIB are good tools for the assessment of older DS. Large multi-centre studies are required to obtain robust, conclusive results. More studies are warranted using any other AD therapeutic approach focused on modifying Aβ deposit and/or neurotransmitter alterations. Donepezil appears to be effective in the treatment of cognitive and behavioral disturbances associated with the progressive dementia syndrome in DS subjects.

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Keywords: Down syndrome, dementia, cognition, Donepezil, PET, clinical assessment.

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One carbon metabolism, Immunology, and Growth hormone in Down syndrome

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Abstract:
The aim of this presentation will be to give a brief update on one carbon metabolism, immunology, and growth hormone in Down syndrome (DS). For the sake of clarity, the three topics will be addressed separately, as follows. One carbon metabolism has been considered in DS, since the pivotal studies of Lejeune (1979) (1). Recent evidence has suggested that an impairment of one carbon metabolism may be a risk factor for DS in the mothers (2), and may modulate the phenotype of people with DS (3). The immunologic derangement in DS consists in a mild immune deficiency, which can be easily counteracted with the use of the available vaccines, antibiotics and the practice of a dynamic life, and the proneness to immune mediated disorders, such as thyroid disorders and celiac disease. Thyroid dysfunction is common in children with DS and warrants the annual screening, but it is uncommon the finding of thyroid antibodies in these children up to the age eight years, when eventually become common. Celiac disease (CD) is the second most important autoimmune disorder of DS. DS is considered an high-risk group for CD, and its screening is recommended by the North American Society for Pediatric Gastroenterology (4). A recent article (5) addresses the cost-effectiveness of screening for CD in asymptomatic children with DS, aiming at preventing lymphoma. The authors maintain that such results do not support the cost-effectiveness of screening. The current and fair conclusion of this debate would be that more data are needed from high-quality randomized clinical trials of screening versus no-screening strategies. Lastly, an involvement of growth hormone in growth retardation and ovarian function in people with DS has been suggested and will be reviewed in the last part of the speech.

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Keywords: One carbon metabolism, immunology, growth hormone, Down syndrome.

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Analysis of Paraoxonase-1, a marker of protection against atherosclerosis in Down syndrome

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Abstract:
Although Down syndrome (DS), one of the most common chromosomal disorders, is associated with a great variety of phenotypes, the incidence of atherosclerotic vascular disease seems to be low (1). The pathogenesis of atherosclerosis is complex and involves multiple genetic and environmental factors. Among the determinants which predispose to premature thromboembolic and atherothrombotic events, the status in the antioxidant enzyme paraoxonase-1 (PON1) is considered as a risk factor for atherosclerotic vascular disease. PON1 is synthesized in the liver and secreted into the serum as an High Density Lipoprotein (HDL)-associated protein which plays a major role in the protective role of HDL against oxidation of low density lipoprotein (LDL) (2). We have recently focused on the interaction between hyperhomocysteinemia and *PON1* expression because hyperhomocysteinemia, characterized by high plasma homocysteine levels, which are often caused by cystathionine beta synthase (CBS) deficiency, is associated with a wide range of clinical manisfestations, like an increased risk for atherosclerosis. Homocysteine, an intermediate thiol-containing amino acid produced by the demethylation of methionine, can be further metabolized into cystathionine by enzymatic condensation with serine by CBS in the transsulfuration pathway. We found a reduced gene expression of *Pon1* in liver of *Cbs* deficient mice, a murine model of hyperhomocysteinemia (3, 4). Since the *CBS* gene is located on chromosome 21, it could be proposed that patients with DS may be protected against atherosclerosis through a phenomenon that is the opposite of that occurring in hyperhomocysteinemia. Then we aimed to analyse the expression of *PON1* in DS fetal liver by quantitative real-time reverse transcriptase-polymerase chain reaction. *PON1* was found to be up-regulated in DS fetal liver (5). Moreover, there was no evidence for an association between PON1 genotypes influencing *PON1* gene expression and DS. Since most serum PON1 is synthesized in the liver, an increase of hepatic *PON1* expression might be one of the factors which could explain the low incidence of atherosclerotic vascular disease in DS. Then our study is carried out in order to investigate the association between the activity of PON1 and circulating oxidized LDL level in patients with DS.

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Keywords: Down syndrome, Paraoxonase-1, liver, gene expression, polymorphisms.

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Autoimmune disorders in persons with Down syndrome

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Abstract:
Introduction: Down syndrome (DS) is associated with an increased risk of immune disorders including autoimmune processes. More frequent disease is Hashimoto´s thyroiditis, Celiac disease, Rheumatic arthritis and autoimmune gastritis. Current recommendations are for regular immune function screening in people with Down syndrome however the significance of borderline results and presence of several antibodies is not clear.
Aim: To examine immune function results of children and adults with DS (aged 0-54) of both sexes follow up regularly in Down syndrome centre in Slovakia
Results: We examined 410 children and 95 adults (aged 0-54) with DS. Of these 5 adults and 2 children had Alopetia totalis and arreata associated with hypothyroidism and positive anti-thyroid, anti-endomyseal and anti-gliadin antibodies. 59% (300/505) of all patients had normal thyroid function, 23,7 % (120/5O5) had a borderline result at their last test and 16,8 % (85/505) had hypothyroidism. Thyroid peroxidase antibody was measured in 200 children and 95 adults. 12,5 % (25/200) children and 40 % (38/95) of adults had a positive test. Celiac disease (positive screening of antibodies AEmA, AGA, aTrg IgA) verified by biopsy were diagnose in 22 children and 30 adults, autoimmune gastritis had 2 patients with DS.
Conclusions: This study confirm the necessity of antibodies evaluation screening in regular examination of persons with DS to prevent the clinical disturbances of autoimmune disorders.

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Keywords: Autoimmune disorders, Down syndrome, antibodies.

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DYRK1A, a chromosome 21 gene involved in central nervous system development with potential phenotypic effect in Down syndrome

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Abstract:
One of the most plausible hypothesis to explain DS is that the extra copy of a few genes in chromosome 21 is involved in determining the phenotype of this complex disorder. One of the genes that have emerged in the last few years as candidate for some of the brain phenotypes associated to DS, including mental retardation, is *DYRK1A*. The reasons that have lead to this hypothesis are the following; i/ the gene is located in the minimal region of chromosome 21 that is considered critical for the syndrome, ii/ it is expressed in tissues that are affected in DS, including the brain; iii/ it is overexpressed in DS tissues; and iv/ transgenic mouse models with moderate levels of DYRK1A overexpression show learning and memory defects (1,2).
DYRK1A encodes a serine/threonine kinase that, through phosphorylation of different substrates such as the *tau* protein or members of the NFAT transcription factors, has the potential to modulate a variety of signalling pathways and cellular processes. By using transgenic mice deficient in DYRK1A protein we have shown that this kinase plays a fundamental role during development regulating growth. *Dyrk1A* null mutant embryos show a severe developmental delay and die during mild-gestation. In agreement with the notion of *DYRK1A* been a dosage-sensitive gene, mice hetererozygous for the mutation, although viable, present developmental delay, a significant reduction in body size and motor alterations. The brain of these mutants is also smaller than normal due to the loss of particular types of neurons (3). In addition, the pyramidal cells of the cerebral cortex of these mice show significant reductions in the length and complexity of their dendrites, thus suggesting the involvement of DYRK1A in neuron differentiation (4). The phenotypic analysis of the retina of mouse models with increased or decreased levels of DYRK1A has recently provided new insights into the role of this protein in CNS development. Still, the molecular mechanisms underlaying the physiological function of DYRK1A are not well known. The challenge now is to uncover which are those mechanisms. This hopefully would help to develop effective therapeutic interventions for DS.

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Keywords: DS, Down syndrome; CNS, central nervous system; NFAT, nuclear factor of activated-T cells.

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Dyrk1a, a therapeutic target?

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Abstract:
A transgenic mouse containing a 500kb human YAC clone (152F7) (Smith et al.) was constructed to model the overexpression of genes from the Down syndrome chromosomal region-1 on HSA21. This model presents two interesting phenotypic modifications: a learning and motor impairment and a volumic increase of some parts of the brain with a more pronounced effect on the thalamic-hypothalamic region as evidenced through MRI experiments.The transgenomic fragment contains five genes among which is dyrk1a, a serine threonine kinase, ortholog of drosophila minibrain. QPCR experiments have shown that the transgene is present in one copy and induces a 1.5 increase of the expression level of dyrk1a in the cerebrum.Crossing of tg152F7 with dyrk1a (+/-) heterozygote produces four genotypes (wt, tg, (+/-), tg (+/-)), analysis of which shows that the brain phenotypes are strongly correlated to dyrk1a gene copy number and to dyrk1a expression level. As a consequence any drug acting upon dyrk1a level or dyrk1a activity should also act upon the phenotype.This kinase has been shown to be strongly inhibited in vitro by epigallocatechin gallate (EGCG) a major component of green tea (Bain et al).We have studied the effect of a PGT diet on the brain volumic increase of YAC transgenic animals as compared to wild type: green tea administered orally during gestation and postnatally can reverse phenotypic changes induced by dyrk1a overexpression.

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Neurogenesis impairment and cell cycle alterations in fetal Down syndrome and Ts65Dn mice brain

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Abstract:

Individuals affected by DS (characterized by trisomy of chromosome 21) display mental retardation and decreased brain volume[1]. Cerebral atrophy is already detectable in fetal and DS children, suggesting that it is due to early developmental defects rather than to late-occurring neurodegenerative processes. To overcome the obstacles inherent in using human material, mice models have been created that replicate, to a different extent, the DS trisomic condition. A DS animal model, the Ts65Dn mouse, shows phenotypic alterations resembling those of DS[2], including atrophy of the hippocampal dentate gyrus[3] and cerebellum[4], two regions decreased in size in DS children and adults[2]. The goal of the present study was to determine whether neurogenesis is reduced during early developmental stages in DS fetuses as well as in Ts65Dn mice, and to identify possible mechanisms underlying this defect. In the hippocampal dentate gyrus and cerebellum of neonate Ts65Dn mice and in the dentate gyrus and ventricular germinal matrix of human DS fetuses, we found that cell proliferation was notably reduced compared to controls. Both DS fetuses and Ts65Dn mice had a higher number of proliferating cells in the G_2 phase of cell cycle, suggesting that their reduced proliferation rate was causally related to a longer time spent in G_2. Consistent with this, microarray screening of the neonatal Ts65Dn mice cerebellum showed decreased expression of genes regulating G_2/M progression. Phenotypic analysis of the surviving cells in Ts65Dn mice showed a reduction in neurogenesis and increase in gliogenesis. These results provide novel insights into developmental derangements in DS and point at cell cycle alterations and differentiation process as critical mechanisms for brain atrophy.

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Keywords: Fetal DS brain, Ts65Dn mouse, neurogenesis, hippocampus, cell cycle.

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Down syndrome and Alzheimer disease

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Abstract:
Down syndrome patients have an abnormal high risk of contracting Alzheimer's disease by the age of 40. The presence of three genomic copies of APP (amyloid precursor potein) that once cleaved generates amyloid-β peptides ultimately forming plaques in the brain of Alzheimer patients is one reason for this high risk. Alternatively large endosomes have been described in neuronal cells of patients with either sporadic Alzheimer's disease or Down syndrome, before the apparition of senile plaques. Since APP is cleaved in this endosomal compartment we suggest that the presence of three copies of APP in neuronal cells with large endosomes could be responsible for the increased amyloid-β peptides formation in Down syndrome patients. We are currently studying APP cleavage in cellular models where the endosomal compartment is modified and looking for genes involved in the large endosome phenotype using mouse models of segmental trisomies.

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siRNA knockdown of the amyloid precursor protein (APP) in the brain *in vivo*

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Abstract:

Trisomy 21, or Down syndrome (DS), is the most frequent genetic cause of mental retardation. The specific molecular cause of DS is unknown. One hypothesis is that increased dosage of a gene or genes on chromosome 21 results in a toxic gain of function causing DS brain abnormalities and cognitive impairment. The cognitive decline and neuropathology in aging DS subjects is strikingly similar to that seen in Alzheimer's disease (AD) patients. Aged DS subjects have profound cerebral atrophy, neurofibrillary tangles, Aβ-containing senile plaques and amyloid angiopathy that are indistinguishable from AD.

Chromosome 21 contains genes coding for proteins involved in brain development and function including the amyloid precursor protein of Aβ, APP (see Kahlem, 2006). While transgenic mice that over express APP clearly indicate the increased APP cannot be the sole cause of DS, it may well contribute to the AD-like neuropathological changes. If this is the case, then reduction of APP in DS may delay cognitive decline and mitigate the severity of the neuropathological changes.

APP is synthesized in the neurons and transported down the axons to the nerve terminal where it is cleaved and rapidly eliminated from the synapse *in vivo* with a half-life of 2-3 hours (Lyckman et al., 1998). We hypothesized that APP plays a fundamental role in normal synaptic function, perhaps by contributing to synaptic integrity, and that changes in presynaptic APP lead to synaptic dysfunction (Moya et al., 2001).

We tested this hypothesis using RNA interference *in vivo* to knockdown presynaptic APP in the brain (Herard et al., 2006). 24 hours after administration, siRNA targeted against APP accumulates in neurons and the APP in axon terminals is significantly reduced. Surprisingly, the amyloid precursor-like protein 2 (APLP2) was reduced as well. Functional imaging experiments in rats during visual stimulation showed that knockdown of presynaptic APP/APLP2 had a profound effect on physiological activity. Namely, APP/APLP2 knockdown significantly decreased stimulation-induced cerebral glucose uptake, a measure of synaptic activation.

The results show that a change the amount of APP delivered to the synapse or in its rate of turnover alters synaptic function. In addition they demonstrate that siRNA can be used to reduce APP in synapses in the brain *in vivo*.

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Keywords: Amyloid precursor protein, axonal transport, synaptic function, RNA interference.

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La spectroscopie de résonance magnétique dans l'exploration métabolique des maladies du cerveau

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Abstract:
La spectroscopie de résonance magnétique (SRM) est une technique d'examen neuroradiologique de plus en plus souvent pratiquée dans l'exploration des pathologies neurologiques en complément des séquences plus classiques d'IRM. Elle permet pour la première fois une exploration noninvasive du métabolisme cérébral basée sur la détection de marqueurs biochimiques des neurones, de la glie et des membranes sans recourir à aucune injection, prélèvement ou radiation ionisante. L'examen par SRM constitue une véritable « biopsie métabolique virtuelle » du cerveau fournissant des informations précises sur plusieurs processus biochimiques définissant l'état métabolique du cerveau normal et pathologique : maturation, souffrance neuronale, altérations membranaires, myélinisation et démyélinisation, activation gliale et gliose, ischémie, invasion macrophagique, etc… Deux techniques de SRM sont applicables en routine à l'animal et à l'homme. La SRM monovoxel analyse un petit volume élémentaire du cerveau dont la localisation est sélectionnée par IRM ; elle est bien adaptée à la caractérisation des lésions focales. L'imagerie métabolique, qui donne la répartition d'un ou plusieurs métabolites sur la totalité d'une coupe en superposition à l'image morphologique classique, s'adresse aux neuropathologies accompagnées d'un désordre métabolique multifocal ou systémique. Les principales indications cliniques de la SRM sont actuellement les processus occupants intracrâniens (en particulier le diagnostic positif d'abcès et de gliomatose cérébrale et le diagnostic différentiel œdème/ infiltration tumorale, la classification des tumeurs…), les encéphalopathies métaboliques d'origine génétique (notamment chez l'enfant, ALD, maladies du peroxisome, du lysosome…), toxique (alcool, encéphalopathie hépatique…) et infectieuse (VIH). Parmi les applications en recherche on peut particulièrement retenir la sclérose en plaques, l'ischémie et la souffrance cérébrale, les épilepsies pharmacorésistantes et les maladies neurodégénératives (Alzheimer…).

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Keywords: MRI, MRS, brain imaging, metabolism, neuropathologies.

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