Abstracts

IX International Symposium on Down Syndrome. Into Old Age: Challenges and Hopes
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Ear, Nose and Throat Conditions and Sleep Disorders

Ear, Nose and Throat Conditions

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Disorders of the upper airways display a number of characteristic features in children and adults with Down syndrome. Typical anatomical traits are short necks, fairly narrow ear canals and nostrils, a relatively backset jawbone (retrognathia) and a somewhat enlarged tongue (macroglossia). This paves the way for specific conditions such as increased mucus production and, particularly, a tendency to snore and to sleep apnea.

Ear disorders are also present at a higher-than-average rate, particularly deformations of the Eustachian tube, which frequently malfunctions in the child's early years. Frequent conditions are otitis serosa or otitis seromucosa, which can lead to conduction hearing loss; enlarged adenoids and/or tonsils, with or without recurring infections; snoring, with or without sleep apnea; chronic rhinitis; and a susceptibility to wax buildup that is conducive to hearing loss. These disorders and their interactions need to be considered, screened for and prevented.

Sleep Disorders

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Sleep disorders in people with Down syndrome (DS) are essentially the same as those affecting the general population. However, except for childhood obstructive sleep apnoea/hypopnoea syndrome (OSAHS), very little specific research has been carried out for DS in this area. OSAHS is best known because of its high prevalence among the population with DS: 30–50% of all subjects, probably rising with age, although adult studies are scarce and insufficiently powered for any conclusions to be drawn. The chief predisposing factor for this syndrome is obesity, but when DS is present other predisposing factors for apnea concur, including a small lower and/or upper jaw, Gothic palate, narrow nasopharyngeal airways, small larynx, tongue protrusion due to a small oral cavity, large adenoids and/or tonsils, increased respiratory secretions, proneness to infection, generalized hypotonia, central nervous system impairments and, occasionally, a subluxated atlantoaxial joint.

OSAHS is caused by a transient total or partial obstruction of the upper airways, which causes pauses in breathing that lead to lower oxygen saturation levels and brief awakenings which make for fragmented sleep. Suspicion of OSAHS is based on certain diurnal and nocturnal clinical manifestations, but diagnosis can only be confirmed with a sleeping polysomnography to establish the type and duration of apnea or hypopnea episodes and their effect on heart rate, blood oxygen and sleep patterns. Treatment must be tailored to each patient and may range from continuous positive airway pressure (CPAP) to surgery.

Another sleep disorder which frequently arises in the teenage years is insomnia, usually linked to psychiatric disorders, particularly anxiety and depression.

Sleep disorders that need to be considered in adults with DS include REM Sleep Behavior Disorder, which frequently arises in patients with Alzheimer-like dementia. However, usually there is just a loss of typical REM atonia without the typical accompanying behavioral disorders.

Finally, there are two syndromes that become more prevalent with age but have rarely been examined in the context of DS: Restless Leg Syndrome and Periodic Limb Movement Disorder, which frequently lead to nocturnal insomnia and, secondarily, to diurnal drowsiness.

Rising life expectancy in people with DS means that research into sleep disorders needs to be extended into adulthood so that treatment can be provided when needed and potential physical and psychological consequences be prevented.

Opening Keynote Speech

The Importance of Medical Care for Adults with Down Syndrome

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During the past three decades we have witnessed numerous studies and read many publications pertaining to the young person with Down syndrome (DS). Although the accumulating literature was welcomed by both parents and professionals, there is limited information available on the maturing and adult person with DS.

This presentation will focus primarily on medical concerns that are more prevalent in adults with DS.

This presentation, focusing primarily on medical concerns that are more prevalent among adults with Down syndrome, highlights the importance of optimal general medical care as well as the intricacies of specific medical issues observed during the maturing years and beyond.

Concerning general health issues we have to ensure that preventative aspects such as up-to-date immunization, nutritional concerns, screening for osteoporosis, hypertension, cancer and others be taken into consideration. With regard to specific medical concerns during adulthood, many issues will be covered, including ophthalmologic, otolaryngologic, respiratory, cardiovascular, gastrointestinal, genitourinary, musculoskeletal, neurological, endocrine, metabolic, psychiatric and other significant health concerns and their treatment in adults with Down syndrome..

In addition, important non-medical issues should not be neglected, including socialization, sexuality, living arrangements, employment, recreation and others that may affect the general well being of a person.

Whereas the majority of adults with Down syndrome will enjoy good health, there are certain general and specific medical concerns that will need attention. Therefore, persons with DS should be examined regularly by their physician and also have laboratory and radiologic tests carried out if needed.

If provided with optimal medical and dental services to foster their well-being in all areas of human functioning, then adults with DS will enjoy a better quality of life and will be able to make a substantial contribution to society.

Moreover, people with DS should enjoy a situation that safeguards their rights and privileges as citizens of a democratic society and effectively preserves their human dignity.

Alzheimer's Disease. Depression. Alarm Triggers and Emotional Support

Dementia and Down Syndrome

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Many articles published over the past 20 years confirm the high prevalence of dementia among people with Down syndrome (DS), whether or not they have mental impairment. Onset is also earlier in this population, partly because of a higher genetic load: it is a well-known fact that chromosome 21 contains the amyloid precursor protein (APP) gene responsible for triggering the «amyloid cascade» widely believed by scientists to be the trigger of Alzheimer's disease (AD) brain lesions.

Pathological studies show the same kind of lesions in the brains of people with DS and those of people with AD. Phenotypically, however, they differ considerably; their different forms of onset and clinical manifestations mean that functional and clinical assessment tests developed for the general population cannot apply in cases of DS

Neurologists specializing in neurodegenerative disorders and accustomed to performing cognitive and functional assessments on AD-type patients often find it difficult to distinguish between prior intellectual impairment and age- or dementia-associated cognitive impairment in patients with DS.

Objective assessment is far more complicated when the patient's direct response to cognitive tasks cannot be evaluated; the physician needs to rely on the subjective accounts of family members, guardians or occupational therapists.

This paper seeks to share an overall perspective on dementia in DS based on the diagnostic and treatment experience acquired at Fundació ACE, and will also make a contribution to cognitive examination and imaging techniques.

Ageing and Depression in Down Syndrome

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There are clearly many different issues that need to be examined regarding ageing in Down syndrome (DS). They include diagnostic difficulties and specific traits of depression in ageing persons with DS, various psychiatric manifestations of Alzheimer's disease, depression as a precursor of dementia, and certain specificities regarding treatment of depression in patients with DS.

Down Syndrome and Dementia: Where Do We Stand and Where Are We Heading?

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People with Down syndrome (DS) are widely known to age prematurely, with a sizeable percentage developing cognitive deterioration.

Early ageing in DS is not synonymous of Alzheimer's disease (AD); neither should every alarm trigger – whether cognitive, behavioral or related to adaptive skills – be attributed to dementia. Differential diagnosis is required to assess cognitive deterioration and consider other kinds of disorders.

There are many frequent causes of deterioration in adults with DS. The acronym DEMENTIA is a helpful memory aid for differential diagnosis: D for depression, E for Environmental changes, M for malignancies – especially gastrointestinal and respiratory with cerebral manifestations –, T for brain trauma and so on.

Each of these variables should be examined from a multidisciplinary whole-person perspective, which is the only way to ascertain what is going on and what the person needs.

Observed behavioral changes often turn out to be a consequence of undetected needs in people with DS, with or without AD.

In fact, when we speak of «alarm triggers and emotional support» we are referring to the need for proper assessment: we must find out what is wrong with the person and what they need. The challenge is how to do it. Concern is visibly rising among health care and social care providers, because of increasing life expectancy among people with DS but also because of our many knowledge gaps.

How can we identify mood disorders and other psychological conditions?

How can we assess whether cognitive deterioration is taking place? What clinical criteria should we use for diagnosis? How can we identify the needs of adults with DS?

It is immediately obvious that appropriate diagnostic instruments are lacking, existing diagnostic criteria are unsuited to the task, staff are underprepared and services inadequately set up to handle cognitive deterioration in adults with DS.

Use of instruments developed for the general population (e.g., MMSE) is utterly mistaken; such instruments have not been validated for this specific population. Materials should be specifically created or adapted for DS in every sphere concerned.

This paper presents some of the instruments currently used in our service to identify alarm triggers (PASS-ADD; CANDID; Test Barcelona-DI; CAMDEX-DS and DC-LD diagnostic criteria) as well as the results obtained in patients with DS.

Health and Hygiene

Health and Hygiene

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Hygiene is defined as the area of medicine devoted to establishing and fostering healthy habits for purposes of disease prevention. It therefore encompasses all the knowledge and techniques that individuals need to apply to control factors that have a direct or potential bearing on health.

The purpose of this discipline is to improve and maintain health and prevent disease.

It is therefore an essential component of preventive medicine.

Developed countries have managed to raise life expectancy through prevention schemes.

People with Down syndrome (DS) have also benefited from social and health-related development, but not on equal terms. For a number of reasons, whether personal, social or cultural, mainstream prevention and screening programs are not easy for them to resort to.

Inappropriate family or social environments largely determine the high prevalence of such issues. The food and hygiene care provided in childhood and adolescence has a direct impact on adult health status; moreover, once the person with DS reaches adulthood, this type of care tends to be gradually and subconsciously neglected by caregivers.

The most prevalent health care issues of adults with DS tend to be more hygiene-, food- and basic everyday-care-related than anything to do with major disorders.

Obesity, orodental disorders and skin conditions, whether isolated or in association, are found to some degree in practically all people with DS.

Obesity

This is the most frequent condition, often underrated by family members who do not worry too much about the physical appearance of people with DS and fail to consider the physical limitations and lower social acceptance linked to obesity.

Guardians need to be made aware of the importance of weight control in DS and an appropriate low-calorie diet needs to be established, maintained and monitored, avoiding impulse eating and setting up regular exercise habits. These are essential basic steps in preventing any type of obesity.

Oral health

Oral health issues are among the most prevalent in people with DS. They begin during childhood because of tooth abnormalities, altered craneofacial development, an impaired immune system, and challenging and hence inadequate oral hygiene.

Skin disorders

Adults with DS usually have dry skin frequently associated with a number of skin disorders that may be hard to treat and cause for concern due to their persistence; better skin care is needed to prevent them.

Osteoporosis

Osteoporosis is a degenerative disorder involving many factors, including protein deficiency, low calcium or vitamin D levels, bed rest, and hormonal disorders such as menopause or hypogonadism.

It is important to implement early preventive measures such as regular daily exercise and a balanced diet including calcium and vitamin D. Dronate treatment may become advisable.

Food and Down Syndrome

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A balanced, appropriate diet is one that meets all of a person's nutritional requirements and thus provides the right amount of energy and nutrients for body functions.

Food intake must be varied and include all types of food. No food is good or bad as such; it all depends on the amount and rate at which it is eaten. People should be encouraged to eat fruit and vegetables, dairy foods, meat and fish, bread and other fiber-rich cereal products, potatoes, and pulses, the latter having been neglected in recent years.

Globally, obesity is on the rise among children and adults alike. This is an even bigger health issue for people with Down syndrome because of their genetic predisposition, although it has dropped somewhat in recent years thanks to preventive care, lifestyle changes and awareness raising among family members.

Parallel to socioeconomic change, recent decades have seen significant change in eating habits and daily physical activity. More animal foods and fats are eaten, to the detriment of fruit, vegetables and pulses, and highly processed foods and beverages are on the rise. This means higher calorie intake with a marked macronutrient imbalance. In addition, processed food is sold in bigger packages to reduce costs, and this leads to excessive consumption of sweets, pastries, snacks and soft drinks.

Another significant development is the drop in physical activity as a result of television watching, sitting around, motor transport, lack of scheduled exercise, and so forth. This is compounded by poor eating habits, and weight gain ensues. Eating and lifestyle habits thus need to be re-taught in the household setting.

The early years are crucial to the learning of good eating

habits; the right patterns should be established at this time to ensure their survival into adulthood.

Nutritional intervention needs to come in early so that obesity and its complications can be prevented. Key concepts on diet and nutrition in both childhood and adulthood hence need to be put across and will play a decisive role in protecting and fostering good health.

Physical Exercise and Down Syndrome

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Motor development in children with Down syndrome (DS) is largely impaired due to musculoskeletal traits such as hypotonia and ligament laxity, which give rise to joint instability including atlantoaxial instability, genu valgum, genu recurvatum, and pes planovalgus among others. These make it difficult to develop good balance and coordination; prevention of biomechanical joint disorders is thus fundamental.

A high rate of obesity is one of the challenges faced by adults with DS. Its etiology may lie with thyroid disorders, a slower basal metabolism, high-calorie food intake, and/or lack of exercise.

Unfortunately, individuals with DS are more likely than average to lead a sedentary lifestyle. Poor eating habits and the other factors listed above compound the effect on obesity. It is therefore important to engage the family in promoting a healthy lifestyle. Inadequate exercise leads to greater health problems, a higher susceptibility to disease and a higher rate of obesity.

Physical activity is beneficial to physical and motor performance, coordination at large as well as fine and gross motor coordination separately, a sense of balance, aerobic and anaerobic performance (endurance, power, etc.), social well-being and quality of life.

One of the challenges faced by our society is finding ways to include people with DS in mainstream sports activities to enhance quality of life and equal opportunities.

People with DS are expected to benefit from practicing sports in a mainstream setting. This should not only improve musculoskeletal strength and general physical condition; it should enhance quality of life at large, improving general well-being, increasing independence and social integration and fostering a better awareness of their limitations, by developing personal habits, independence, relational skills and personal skills, among others. Taking part in sports helps individuals with DS feel that they belong and affords opportunities to test and discover their own capabilities, reach individual decisions and achieve a higher degree of independence.

Sexuality: Medical, Psychological and Legal Issues

Sexuality: Medical Issues

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Sexuality, as a form of expression within human behavior, is part of life for people with disabilities, and thus for people with Down syndrome (DS), just as it is for the rest of humankind.

This natural bent needs to be educated, just as education is required for all other elements of social and personal relationships hence included in school curricula. Transmission of sexual values must be therefore be part of education as a whole.

Sexuality should be portrayed as normal, with a particular emphasis on issues such as privacy, respect, dignity and so forth.

However, this has consequences that need to be assessed and analyzed from different perspectives: sexual expression, reproduction, contraception, sterilization, sexually transmitted diseases, sexual abuse, and so forth – concerns which are also raised with teenagers at large. This requires an exercise in individual and collective reflection in order to provide information, show respect, and discuss and consider all the different options so that the people with disabilities can make appropriate decisions and their social circles, particularly parents but also caregivers and professionals, can dispel anxieties.

This requires a joint commitment by everyone involved, both to contribute appropriate input, for the sake of the individual's dignity, and to simplify it adequately, so that the different choices do not become an insurmountable barrier but rather a trail that can be feasibly followed and shared by the individual, with the help of the parents and other stakeholders, in a way that is suited to individual development, and in compliance with the law.

Sexuality: Psychological Issues

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Sexuality is just one more dimension in the overall development of human beings. We all have a sexual body and our sexual-affective needs play a role in our relationships throughout our lives.

The sexuality of individuals with disabilities is a topic of concern which arouses some interest and is surrounded by prejudice. Adults with disabilities have difficult social and affective relationships, including difficulty in achieving a rewarding sexual life, but this is not solely due to disability itself: in part it is because of difficulties in finding their own true identity and constructing their own sexuality. Sexuality is an adult practice; thus, in order to lead satisfactory sex lives, they need to become adults.

While there are indeed many prejudices regarding the sexuality of individuals with disabilities, it is in fact no different from human sexuality at large, and displays all three dimensions: reproduction, pleasure, and affection.

Sexuality is not only genital. It is about the ability to grow into a relationship that integrates different aspects: falling in love, attraction, desire, protection, sharing an ongoing project. An understanding of how an individual with a disability integrates all these can pave the way for the utmost possible normalcy. Their sexuality needs to be acknowledged, chronological age has to be taken into account, the individual has to be viewed as an evolving being, and education has to encourage the completion of each stage in sexual development without getting stuck at any particular level, while information is supplied and trust is conveyed.

To educate a person is to convey rules, but also to allow them to grow. A person with a disability needs help to integrate functions, emphasizing those where the person is farther ahead so that every aspect in their personality can be fully developed.

Legal Aspects
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Disability of any sort does not necessarily deprive an individual of any rights or duties. However, it is known to make a person more vulnerable in many cases. It is therefore important to know what can be done to ensure that everyone can exercise their rights and fulfill their duties equally on a lifelong basis.

Sexuality, as an inherent part of a person's being, may have legal consequences which, if known and taken into account, may safeguard the correct and full exercise of the person's rights.

The panel on «Sexuality: medical, psychological and legal aspects» held within the IX International Symposium on Down Syndrome was intended as an opportunity to reflect, among other subjects, upon a number of sexuality-related legal issues that turn out to have an impact on people with some sort of psychic disability, including:

– Guardians and sexuality. A reference to the legal protection of people with disabilities: total or partial legal incapacitation and the appointment of a guardian.
– Court procedures and key concepts regarding sterilization.
– Awareness of how sexual abuse is treated under criminal law as well as the circumstances attending upon criminal proceedings.

Other legal matters, though not as directly linked to sexuality, may impinge on the individual with Down syndrome, including:

– Future planning actions by a person with a psychic disability: special powers of attorney, living wills and self-guardianship.
– Awareness of Spain's new Dependency Act and Catalonia's Social Service Act.
– Awareness of certain Social Security-related matters.

Community Commitment to Social Inclusion

Community Planning and Municipal Engagement in the Lives of People with Disabilities and Their Families
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Community based services and supports for people with disabilities and their families continue to evolve. Today, services and supports, although increasingly community-based, operate within a formal system outside of the community development and decision-making structures. This may, in part, explain the relative exclusion of persons with significant disabilities from community life, integrated employment, and political engagement. Research and practices related to community development, public health, social capital, grass-roots organizing, universal design, social capital, and consumer-directed services and supports can provide a useful foundation for advancing beyond these current community-based systems. A recent demonstration project in a small rural community in New Hampshire illustrates the potential of organized communities to develop local support structures and mechanisms that result in enhanced quality of life and social integration for people with disabilities and their families.

Despite the fact that over time, the community service system has modified expenditure patterns in favor of smaller and more individualized housing and employment options, family support and respite care, and most recently, consumer driven services (CMS, 2004) it has retained a basic organization that is fundamentally exceptionalistic. In response to calls for consumers to be more closely connected to their communities, a number of innovative practices have been developed, including employing community connecters and «bridge-builders» (McKnight, 1998), personal futures planning (Mount, 1987; O'Brien & Mount, 1991), engaging citizens as advocates and informal guardians (Herr, 2001), and self-determination and individualized budgeting (Nerney & Shumway, 1996).

In response to these social realities, John McKnight (1995) and his colleagues have been outspoken critics of the currently constructed human service system. They argue that the system has served to alienate individuals with disabilities from their communities, and disrupt the natural role that communities play in the lives of individuals. This may, in part, be due to a focus on exceptionalistic services that are disconnected from the local community infrastructures that impact the day to day community expeexperience of individuals with disabilities, such as housing stock, jobs, transportation, recreation, education, and so on.

What is now needed is an overall conceptualization of how local communities can be systematically engaged to play a central role in developing community-centered «solutions that recognize individuals with disabilities as part of the community. The municipal infrastructures that impact housing stock, jobs, transportation, and physical accessibility in the U.S. include Planning Boards, Zoning Boards of Adjustments, and Town Councils. Each has a role in addressing long-term care services and supports. Access to affordable and accessible housing is an example of an area where communities can have an impact. Although Housing Finance Agencies have the ability to leverage dollars to support the production of housing, communities decide what is built through local Master Planning and Zoning Ordinances. When housing developers request a permit to build multiple units, a community can ask, in return for local variances and waivers, that a certain number of affordable and accessible units be built. Housing developers can get support to do this though local housing agencies or the Housing Finance Authority. Similarly, transportation solutions can be developed locally or regionally through regional planning commissions. Decisions as to the purchase of accessible vehicles that serve the public are not made by service providers but, rather, through regional groups with local representation. Accessible recreation facilities are made available to all community members including those who have disabilities and/or are aging. Finally, locally organized solutions for people with disabilities could address a variety of other issues such as chronic underemployment and unemployment. Like the Genoa, Italy Model (Gerry, 1999), local groups of business leaders, not dissimilar to Rotarians, with support from «employment facilitators» (Sowers, Milliken, Cotton, Sousa, Dwyer, & Kouwenhoven, 2000) support young people with disabilities graduating from high school as well as others in need of productive employment. In essence, we are striving for livable communities for all people. This cannot be achieved without the involvement of municipal and civic leaders and planning structures.

Independent Living and Social Inclusion
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When we speak of independent living we are referring to people with intellectual disabilities who either intentionally or as a result of circumstances live in a home of their own and manage their daily lives, with support provided to foster their well-being. It is an excellent chance to analyze the quality and quantity of opportunities and obstacles for the social participation of these individuals in their own natural settings.

According to Dr. Rober Shalock, «Community is the context for quality of life». This statement clearly and roundly sets the horizon of the aims and challenges taken on by professionals and service providers when defining our task in regard to the individual with a disability.

From the perspective of a service providing support to individuals through the process of achieving independence, our experience affords an analysis of how the individual and his or her social environment are adapting mutually, the kind of natural support the person enjoys when shopping, requesting a service or seeking a solution to a specific need, what kind of network he or she can draw upon, how interpersonal relationships are managed, what degree of diversity is present in their setting, how the community is facilitating accessibility, and so on.

Similarly, an awareness of the focus person's aspirations as regards access to and enjoyment of the community can usefully help decide which particular areas require support in the planning process. The role of service providers in this regard will also need to be analyzed, as well as the thrashing out of decisions between the individual's self-determination and the will expressed by his or her reference persons.