Specific aspects of ageing in Down’s syndrome

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Abstract

Life expectancy is increasing considerably in the population with Down’s Syndrome (DS), thanks to advances in health care and living conditions. As a result, older people with DS require specialized care. This article aims to give an updated view of ageing and end of life in people with DS, with three sections: 1. Definition of geriatric concepts. 2. Specific medical problems of ageing in DS. 3. End of life in DS. Finally, a set of guidelines for the detection of specific problems in the care of older patients with SD is proposed.

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PALABRAS CLAVE
Síndrome de Down; Geriatría; Envejecimiento; Fragilidad; Mortalidad

Aspects específicos del envejecimiento en el síndrome de Down

Resumen

La esperanza de vida está aumentando considerablemente en la población con síndrome de Down (SD), gracias a los avances sanitarios y a las condiciones de vida. Como consecuencia de este fenómeno, surgen necesidades específicas para las personas con SD de edad avanzada, que requieren una atención sanitaria especializada. El presente artículo pretende dar una visión actualizada del envejecimiento y final de vida en la población con SD, mediante tres apartados: a) definición de conceptos geriátricos; b) problemas médicos específicos del envejecimiento en el SD, y c) final de vida en el SD. En último lugar, se propone una guía de recomendaciones para detectar problemas específicos en la atención de pacientes con SD de edad avanzada.

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Introduction

The increase in life expectancy of the general population in the last decades has led to complex clinical procedures. Ageing involves changes in physical and cognitive capacities, which are generally accompanied by changes in the family and social environment. At physiological level, it is considered a multifactorial process, with a common denominator: chronic and progressive inflammation which affects multiple physiological systems. Ageing is an individual process, and chronological age may not reflect the functional reserve or the life expectancy of a person.1

Due to advances in health care and living conditions, the increase in life expectancy is now being recognized in the population with Down's syndrome (DS), although this ageing process at begins at an earlier age, with a decrease in DNA repair ability, increased biological age, and earlier death.2

According to the medical literature, early ageing in people with DS starts around 45 years of age, with an increased risk of particular health problems and functional and cognitive impairment.3 Some of the health factors that have contributed to the increase in life expectancy in this group have been, the correction of congenital heart diseases, development of immunisation programmes and approaches to infections, improvements in nutrition and specific health problems. Among the social factors are highlighted, less institutionalisation, and the introduction of specialised community health services.4 It is surprising that in 1900 the mean life expectancy was 9-11 years, while it is currently greater than 50 years, and some even reach 70 years.5,6 It is estimated that the mean life expectancy will reach 60 years in future generations.2

As a result of this phenomenon, some specific needs, which require specialised care, are emerging for people of advanced age with DS. The present article attempts to give an updated view of ageing and end of life in the population with DS, divided into three sections: 1. Definition of geriatric concepts; 2. Specific medical problems of ageing in DS; 3. End of life in DS. Finally, taking these three sections into account, a set of guidelines are proposed for detecting specific problems in the care of elderly DS patients.

Definition of geriatric concepts

Loss of functional capacity7,8

Functional capacity is the ability a person has to carry out an activity by him or herself. The loss of functional capacity gradually progresses to functional impairment, physical disability (difficulty in carrying out activities of daily life [ADL]), and dependence (the need for assistance by other people to carry out ADL). It is well known that life expectancy not only depends on age and comorbidity, but also that an important predictive factor is the functional situation.

Physical functional capacity in the general population is mainly measured by ADL, which differentiate between basic activities of daily living (BADL) and instrumental activities of daily living (IADL). The BADL mostly consist of personal hygiene, on the ability to go to the toilet, to be able to dress oneself, eat, mobility, and sphincter control. It is usually measured using the Barthel index, and changes in this indicate the need for a caregiver. The IADL are more complex needs required for the independent life, and consist of the ability to be responsible for medication, shopping, money, telephone and public transport. It is mainly measured in the general population using the Lawton index. Scales such as the Pfeiffer index are used for measuring mental status.

Numerous studies have shown that there is a more accelerated functional and cognitive impairment in DS from 50 years of age than in the general population, and at earlier ages and with a greater risk of Alzheimer's disease.3,4 However, there is no higher risk of functional impairment at ages less than 40-50 years, and people with DS even have better abilities than other types of mental retardation.3

There is a need to have tools available adapted for this group, as well as specific training in this field, both as a guide to diagnosis and for intervention.4 A modified Barthel index functional scale, adapted for people of advanced age with mental retardation, has been used in some studies. It is a scale with 30 items that measure basic, as well as instrumental abilities, in areas such as housework, personal hygiene, food and mobility.3

With the objective of homogenising scales for measuring cognitive abilities in DS, an International Working Group for the diagnosis of dementia in people with intellectual disability proposed an assessment battery applicable for the majority of adults with intellectual disability. This includes, on the one hand, questions directed at the caregiver (“Dementia Scale for Down's Syndrome [DSDS], "Dementia Questionnaire for mentally retarded persons [DMR]"), as well as questionnaires administered to the patient (CAMCOG-DS, "Test for severe impairment", and other specific neuropsychological tests.6

Comprehensive geriatric assessment1

An comprehensive geriatric assessment consists of an interdisciplinary diagnostic process directed at identifying and evaluating multiple dimensions of the person; medical problems and comorbidity, functional capacity, physical, mental (cognitive, emotional) and social functions (Fig. 1.) It is an exhaustive and protocolised assessment with standardised scales. Its purpose is not only to restore health, but also to maintain as high a level of independence as possible, as well as mental and physical independence, thus achieving a better quality of life in the ageing process. The comprehensive assessment must be followed by a therapeutic plan with multifactorial interventions.
Frailty9-12

One of the most accepted definitions of frailty is: “An increased physiological state of vulnerability to stress factors as a result of a decrease or deregulation of physiological reserves of multiple biological systems that give rise to difficulty in maintaining homeostasis”.9 It is a biological syndrome of reduced functional reserve, and thus leads to progressive functional impairment and finally, death.

Specific frailty indicators related to ageing and dementia need to be devised for the population group with DS.11

Comorbidity and compression of comorbidity

Comorbidity implies the presence of several disorders or diseases. The fact of delaying comorbidity implies a delay in disability and dependence, as such that it increases active life expectancy. In this sense, they are essential preventive measures. A good measure of health status is the disability-free life expectancy, as it takes into account the duration of life, as well as its quality, estimated by functional limitation. Minimising the progression of disability is of great interest in patients with DS. The aim is not just to prolong life, but also “to live it”, with more health and independence, “to compress” the greater disability and comorbidity towards the end of life.13

Comorbidity, frailty and disability are three overlapping conditions,10,12 but they have their differences. As mentioned previously, comorbidity implies the presence of various diseases: disability implies restriction or loss of the ability to carry out an activity; and frailty, vulnerability and loss of homeostasis due to a decrease in functional reserve. Frailty and comorbidity are predictors of disability, and thus, can also exacerbate comorbidity and frailty.

In DS, the presence of comorbidities is increased around 45 years of age, with an increased risk of specific health problems, and functional and cognitive impairment.3

Specific medical problems of ageing in DS

People with DS at advanced ages (>45-50 years) have a higher risk of dementia, dermatological changes, early menopause, sight and hearing disorders, late onset epilepsy, hypothyroidism, obesity, sleep apnoea, osteoporosis and other musculoskeletal diseases, limitations in cardiorespiratory capacity, and a higher susceptibility to infections. On the other hand, it should be pointed out that there is a lower prevalence of other diseases such as, solid malignant tumours, hypertension, coronary and cerebrovascular diseases. Table 1 shows the differences in the principal diseases by age groups.

The most common diseases that elderly people with DS suffer are reviewed next.2,6,14-17

Dementia15

Alzheimer’s disease is the type of dementia most associated with DS, as it occurs more often and at an earlier age than in the general population. The extra chromosome 21 leads to the production of beta-amyloid protein, which gives rise to the deposit of the amyloid plaques in the brain associated with Alzheimer’s disease. In post-mortem studies, it has been observed that approximately 80% of people over 40 years-old develop the neuropathological characteristics of Alzheimer’s disease, although the appearance of clinical symptoms may occur some years later.2 It appears that 8% of the population group between 35-49 years, 55% between 50 and 59 years, and 75% of over 60 year-olds, develop dementia.4 With the research currently carried out in this field, the percentage diagnosed is probably higher than in previous studies. Another type of less frequent dementia, but has also been detected in people with DS, is dementia due to Lewy bodies.4
Detecting the first clinical signs of dementia is complex, due to the difficulty in distinguishing it from previous cognitive deficits, a depression, delirium, or hypothyroidism. The presence of sensory deficits may also mask the symptoms. A stressful life event may often be the triggering factor of the onset of Alzheimer’s disease.

The diagnostic criteria of dementia must be taken into account:

— Deterioration from the previous short and long-term memory level.
— Involvement of at least one other cognitive function, that may be manifested as one of the following symptoms: change in the baseline level of temporal and spatial orientation, aphasia, progressive loss of acquired oral and written language, apraxia, and loss of manipulative abilities.

To be able to diagnose dementia, these two criteria must be accompanied by an inability to carry out normal daily activities, that is to say, a change in social and/or work function, or in the level of independence in activities of daily living (basic or instrumental). In the more advanced phases behavioural disorders may appear, such as changes in personality, changes in perception, delirious ideas, aggressive behaviours, lack of inhibition (inappropriate sexual or social behaviour), irritability, apathy, or repetitive behaviours. Neurological signs, such as urinary or faecal incontinence, changes in gait, tremor, and rigidity, usually appear in the moderate to severe phases. A good diagnosis requires good knowledge of the previous functioning, both cognitive and functional, of the patient.

**Sight disorders**

Sight problems (44-71%) are common in DS of advanced age, and in a large percentage of the general population. It is important to mention, that sight disorders have been under-diagnosed in the studies conducted, and as a result these problems have not been adequately corrected. Among the most frequent ophthalmic problems are cataracts, strabismus, refraction problems, and keratoconus. Sight disorders may persist even when surgery is performed, due to aphakia and the poor use of bifocal lenses.

**Hearing disorders**

Age-associated hearing loss is more common in DS of advanced age, and in a large percentage of the general population. Neurosensory high-frequency hearing disorders start about 30 years before that in the general population, with a more rapid and progressive degenerative process. Up to 70% of adults with DS from 40-50 years have hearing loss, with 53% moderate and 17% severe. It is also important to highlight that hearing disorders are often under-diagnosed or detected late, and that correction with hearing aids is lower than expected.
loss causes a decrease in sensory afference, with difficulty in detecting verbal information in the environment, leading to social isolation and a lack of cognitive stimulation.

Another aspect that needs to be taken into account is the problem of cerumen (ear wax), which easily obstructs the external ear canal, on being narrower than in the general population. It is important to follow this up and remove it, as it could make the hearing loss worse.

Dermatological changes

Characteristic symptoms of premature ageing appear in adults with DS, such as premature greying of the hair, alopecia areata and wrinkling of the skin. Other skin problems are, atopic dermatitis, fungal infections, seborrheic dermatitis, and xerosis. Some of these disorders may be partly due to the decline in self-care associated with memory loss and dementia.

Convulsive disorders

The frequency of seizures increases with age in DS, being 24-28% in adults over 50 years. The type of seizure suffered is generally tonic-clonic, or simple or complex partial seizures. The late onset of epilepsy also seems to be associated with the start of dementia (75-85% of older DS adults with Alzheimer’s disease).

Thyroid dysfunction

The prevalence of thyroid disorders in adults with DS increases with age, with 35-40% of adults having abnormal thyroid function, and 7-8% with active hypothyroidism. It is important to note that it can be a treatable cause of cognitive impairment. The need to systematically review thyroid function is well known.

Obesity

The frequency of overweight and obesity in adults with DS is greater than 50%, which carries cardiovascular (arterial hypertension), osteoarticular, and metabolic risks (dyslipidaemia, diabetes, hyperuricaemia). Among the factors responsible are, a lower level of physical activity, eating habits, basal metabolism, and hypothyroidism.

Sleep apnoea syndrome

Adults with DS have a higher prevalence of obstructive sleep apnoea (as high as 60%), due to the morphological characteristics that come with ageing (larger tonsils, glossoptosis, and other facial and respiratory tract factors), and is also linked to obesity.

Musculoskeletal diseases

Osteoarticular problems are often a result of premature degeneration of bones, and joint disease. The risk of osteoporosis is higher in adults with DS, and increases with, age, as a result of early menopause, less physical activity, or low muscle tone. Degenerative osteoarthritis is also common, which presents with symptoms of numbness, weakness, and pain. Orthopaedic problems are frequent, and associated with congenital osteoarticular anomalies, such as flat feet.

Atlantoaxial instability in the spinal column may also be present in DS adults of advanced age. Although it is uncommon (1.5%), it may produce neurological symptoms such as, difficulty in walking, neck pain, or torticollis. Other rare degenerative skeletal disorders are dislocated hip and partial dislocation of the kneecap.

Mitral valve prolapse

It is more common in adults with DS (46-57%), and its delayed appearance is independent of congenital heart disease. It is important to record antibiotic prophylaxis in dental check-ups.

Early menopause

Women with DS start menopause at a mean age of between 45-47 years, some 4-6 years earlier than in women with other intellectual disabilities, and the general population. This involves a higher risk of heart diseases, osteoporosis and breast cancer.

Infections

The change in humoral and cell-mediated immunity of the population with DS may lead to a higher risk of infectious diseases. Pneumonia is common in DS adults of advanced age, and is the main cause of death in this group. Aspiration pneumonias are also a complication of the specific morphological characteristics of DS, including swallowing disorders due to the decrease in structural flexibility with age, oesophageal and pharyngeal peristalsis, pharyngeal sensitivity (decreases the detection of liquids and the food bolus, and the contents of the pharynx may pass into the trachea). The depressed immunity may also lead to oral, dental and dermatological disease, thus it lends special importance to oral hygiene and skin care.

Psychiatric disorders

Depression is the most frequent psychiatric disorder in the population with DS, and ageing also increases the incidence, and is more prevalent than in the general population, or in other types of mental disorder. Some triggering factors are, loss of sight or hearing, loss of functional capacity, and death of a close member of the family or caregiver. Like dementia, a diagnosis of depression is difficult to establish in DS, mainly due to the difficulties in expressing and communicating their emotional state. Some symptoms that
they may have are: sadness, affective lability, tendency to isolation, apathy, lack of energy, mood changes, somatic symptoms, fear, etc. Psychotic symptoms, such as changes in behaviour and thoughts are also relatively frequent. The psychotic symptoms and somatisation are more common in older people with DS than in younger people, and anxiety and crying are higher.

The prevalence of depressive symptoms in people with Alzheimer’s disease (20%) is much higher than in the general population. The differential diagnosis between dementia and depression can be difficult (they can also be present simultaneously). Depression is more fluctuating, reversible, and generally responds well to treatment. Apathy is the most frequent neuro-psychiatric symptom of onset of Alzheimer’s disease.

End of life in Down’s syndrome

In the general population there are four end of life trajectories: 1. Sudden death (5%); 2. Advanced chronic organ failure (cardiac, renal, respiratory, hepatic) (30%); 3. Cancer (25%), and 4. Dementia and other neurodegenerative diseases (40%).20,21 A terminal disease is defined as an advanced, progressive disease, with little or no possibilities of response to specific treatments, and that leads to a prognosis of weeks or months of life.

In DS there is a different mortality pattern to that of the general population. In a recent study, the cause of death of 97 persons over 40 years-old with DS was determined, and it was found that pneumonia and other respiratory infections were the main cause of death (40%), followed by coronary disease (30%), heart, renal and respiratory failure (9%), and cerebrovascular accident (5%). The factors that better predict mortality are: age, functional deterioration, and behavioural changes.20,21

The palliative care needs in the last stage of life should be the same as in the general population. There is a need to integrate palliative care skills and knowledge of people with disabilities, in order to provide a better quality end of life.20,21

In the elderly with DS and comorbidity, functional deterioration, dementia, and any other serious illness that has a poor short-medium term prognosis, it is recommended to document the symptoms, concerns of the patient, the situation of the caregivers, to approach the practical problems and begin advanced end-of-life planning. These aspects can be mentioned in a living will document. In the end-of-life situation it is recommended to prioritise comfort and symptom control due to medical complications, avoid aggressive treatment as well as under-treatment, attempting to carry out a diagnostic-therapeutic approach proportionate to the baseline physical and cognitive situation of the patient. “Good death” is understood as that which occurs free of avoidable suffering, for the patients and their families, respecting the wishes of the patient.21

Recommended guidelines for the detection of specific problems in the care of the elderly patient with Down’s syndrome

The recommendations for achieving healthy ageing in people with DS, and concluded from the previous sections of the article, are set out in Table 2.

The comprehensive geriatric assessment should include an evaluation of medical problems (a check-list of the most common diseases, symptoms, and medication), as well as physical, cognitive, and social functional capacity. It is recommended to perform the following laboratory tests periodically: blood glucose, total, LDL, and HDL cholesterol, zinc, TSH and free T4, folic acid, vitamin B12, calcium and vitamin D, as well as the usual routine analysis parameters.

Conclusions

The longer life expectancy of people with DS, which already reaches 60 years, brings with it a higher prevalence of comorbidity associate with ageing. It is often difficult to differentiate between some diseases (dementia, depression, etc.) of “normal” ageing.2 In a study conducted on the quality of life, it was shown that people over 45 years of age with DS are considered young or middle aged, they feel happy, and give much value to their privacy, and they see themselves as adults.5

The clinical approach to ageing in DS requires a multidisciplinary medical team (geriatrics, family doctors, and other specialties), psychologists, physiotherapists and nursing staff, who work together, and with specific training in this field. It is also necessary to have suitable and standardised tools available. The common objective is to improve, the prevention, the diagnosis, and therapeutic options in order to improve their independence and quality of life at the end of life.4,15 Furthermore, society should make it easier to have access to the services, healthcare levels, and pharmacological treatment under the same conditions as the rest of the population.4

Research is needed in order to gain better knowledge of the natural history of the medical diseases in the ageing process of people with DS, to improve the preventive approach and specific treatment, and to provide them with the maximum well-being in their old age.

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Conflict of Interests

The author declares not to have any conflict of interests.
Table 2  Principal medical problems to take into account in assessing patients of advanced age with Down’s syndrome

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<th>Recommendations</th>
<th>Observations</th>
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<td>Hearing disorders</td>
<td>Removal of ear-wax plugs</td>
<td>Early detection and correction prevents social isolation and loss of cognitive performance</td>
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<td></td>
<td>Early detection of presbycusis</td>
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<td></td>
<td>Periodical ear, nose and throat check-ups</td>
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<td>Sight disorders</td>
<td>Periodic reviews by ophthalmologist</td>
<td>Early detection, particularly cataracts</td>
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<tr>
<td>Sleep apnoea syndromes</td>
<td>If suspected, refer to a specialist</td>
<td>Presence of nocturnal apnoeas, number of apnoeas/hour (5/hour could be pathological)</td>
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<tr>
<td>Oropharyngeal dysphagia</td>
<td>Changes in the consistency of the diet to prevent pneumonias due to bronchoaspiration</td>
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<tr>
<td>Thyroid disorders</td>
<td>Periodic reviews of thyroid hormones (1-2/year)</td>
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<tr>
<td>Seizures</td>
<td>Ensure antiepileptic treatment</td>
<td>Presence, type and frequency</td>
</tr>
<tr>
<td>Heart disease</td>
<td>Review mitral insufficiency systolic murmur, heart failure symptoms, arrhythmias, syncope</td>
<td>Control blood pressure and glucose</td>
</tr>
<tr>
<td>Orthopaedic disease</td>
<td>Rule out foot problems, atlantoaxial instability symptoms, or other diseases. Ensure good fitting footwear</td>
<td>Assess radiological study and referral to orthopaedics</td>
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<tr>
<td>Osteoporosis</td>
<td>Ensure a calcium and vitamin D rich diet, physical exercise</td>
<td>Review whether specific treatment is required</td>
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<td>Sphincter incontinence</td>
<td>Detection and preventive recommendations</td>
<td>Types: urinary, faecal</td>
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<tr>
<td>Depression and anxiety</td>
<td>Detection of symptoms and therapeutic evaluation</td>
<td></td>
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<tr>
<td>Dementia</td>
<td>Rule out reversible causes like hypothyroidism, folic acid/ vitamin B&lt;sub&gt;12&lt;/sub&gt; deficiency, depression, delirium or sensory deficits If there is suspicion, request a neuropsychological assessment to carry out an evaluation with tools specifically designed for DS, and refer to medical specialist in dementia (neurologist/psychiatrist/geriatrician) to assess neuroimaging studies and specific pharmacological treatment. In a population with intellectual disability, it is recommended to perform at least one neuropsychological assessment before 30 years of age, to determine the baseline status of the patient</td>
<td>Dementia criteria: memory disorder, involvement of at least one other cognitive function, and interference in activities of daily living. If death, assess the neuropathological study of the brain tissue to progress in the research</td>
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<tr>
<td>Polypharmacy</td>
<td>Review drugs, therapeutic suitability and interactions</td>
<td>Ensure therapeutic compliance</td>
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<tr>
<td>End of life</td>
<td>Identify end-of-life situations, resolve practical aspects and base the decision making on prioritising comfort</td>
<td>Caregivers and family should know the alert symptoms and have instructions on what to do</td>
</tr>
<tr>
<td>Functional status</td>
<td>Previous and current physical and cognitive functional capacity. Assess whether there has been a deterioration in the last weeks/months Ask about the social and work activity situation. Identify the caregiver and guardian</td>
<td>Assess any functional scale specific for DS (e.g. modified scale of the Barthel index)</td>
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(Continue on next page)
Table 2  Principal medical problems to take into account in assessing patients of advanced age with Down’s syndrome (continuation)

<table>
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<tr>
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<th>Recommendations</th>
<th>Observations</th>
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<tbody>
<tr>
<td>Important general recommendations</td>
<td>Physical exercise, to prevent obesity, osteoarticular rigidity, osteoporosis, falls. Help to maintain the independence, improve the emotional state, and increase the overall quality of life. Healthy eating habits: balanced, varied and fractionated diet. If there is obesity, low calorie diet, maintained and controlled, and maintain regular physical exercise. Oral hygiene. Periodical dental check-ups (once/year). Record antibiotic prophylaxis if there is mitral valve prolapse. Care of the skin (daily hygiene, moisturising cream). Cognitive stimulation activities. Help and improve social activity.</td>
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<tr>
<td>Updated preventive vaccinations</td>
<td>Tetanus toxoid vaccine, pneumococcal vaccine and flu vaccine</td>
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References