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Italian Guidelines for the diagnosis and treatment of Fetal Alcohol Spectrum Disorders

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Summary. Drinking alcohol during pregnancy can cause congenital disabilities. J. Roquette, P. Lemoine and K.L. Jones were the first to describe these effects. In 1973, Jones and Smith coined the term Fetal Alcohol Syndrome to describe children with facial anomalies, poor growth, and learning difficulties. The caution against drinking during pregnancy has existed for centuries, including in The Bible (Judges 13:3-4). Maternal alcohol consumption is linked to congenital disabilities. To ensure safety, it is advised to abstain from alcohol during pregnancy. Fetal Alcohol Spectrum Disorder (FASD) was observed in paintings from the mid-19th century when artists began depicting moments and characters from everyday life. In 2005-2006, Italy conducted a groundbreaking study on FASD, the first in Europe. The study resulted in valuable research on FASD, contributing to prevention efforts. Unfortunately, diagnosing FASD remains a challenge in Italy. Early diagnosis and treatment are critical, and increasing the number of authorized centers to diagnose FASD is necessary to improve care. Educating ourselves about FASD is the key to creating a world where affected children receive the care they need. These guidelines include nine works dealing with all FASD aspects such as prevention, the effects on cognition, the epidemiology, the diagnostic criteria, the clinical aspects, the general effects on the body, the available treatments and the methods of detecting alcohol abuse in pregnant women.

Key words. Alcohol, disability, FASD, gestation, pregnancy.

Linee guida italiane per la diagnosi e il trattamento dei disturbi dello spettro feto-alcolico.

Riassunto. Bere alcol durante la gravidanza può causare disabilità congenite. J. Roquette, P. Lemoine e K.L. Jones furono i primi a descrivere questi effetti. Nel 1973, Jones e Smith coniarono il termine sindrome alcolica fetale per descrivere i bambini con anomalie facciali, scarsa crescita e difficoltà di apprendimento. Una certa cautela nel bere durante la gravidanza esiste da secoli, come riportato anche nella Bibbia (Giudici 13:3-4). Il consumo materno di alcol può determinare l'insorgenza di disabilità congenite. Per garantire la sicurezza, viene consigliato di astenersi dall'alcol durante la gravidanza e l'allattamento. Una testimonianza del disturbo dello spettro feto-alcolico (FASD) è possibile osservarla anche nei dipinti della metà del XIX secolo, quando gli artisti iniziarono a rappresentare momenti e personaggi della vita quotidiana. Nel 2005-2006, l'Italia ha condotto uno studio innovativo sulla FASD, il primo di questo tipo in Europa. Lo studio ha prodotto preziose ricerche sulla FASD, contribuendo agli sforzi di prevenzione. Sfortunatamente, diagnosticare la FASD rimane una sfida in Italia. La diagnosi e il trattamento precoci sono fondamentali e per migliorare l'assistenza è necessario aumentare il numero di centri autorizzati per diagnosticare la FASD. Queste linee guida comprendono nove lavori che trattano tutti gli aspetti della FASD come la prevenzione, gli effetti sulla cognizione, l'epidemiologia, i criteri diagnostici, gli aspetti clinici, gli effetti generali sull'organismo, i trattamenti disponibili e i metodi di rilevamento dell'abuso di alcol in gravidanza.

Parole chiave. Alcol, disabilità, FASD, gestazione, gravidanza.

In the Modern Era, the first to describe the symptoms and clinical signs of the effects of alcohol on the fetus and its development were J. Roquette[1], P. Lemoine and K.L. Jones[1,2]. The term Fetal Alcohol Syndrome (FAS) was used for the first time by Jones and Smith in 1973: in the publications, the author describes a group of children born to "alcoholic" mothers who had characteristic facial anomalies, poor prenatal and postnatal growth, and neuro-developmental and learning problems[3]. However, the deleterious and teratogenic effects of alcohol on the developing fetus have been known for centuries. The first references to the association of alcohol/fetal malformations are found in the Greek and Roman traditions, according to which alcohol intoxication, at the time of procreation, causes the birth of a child with congenital disabilities.

Historical records show that knowledge of the adverse effects of drinking during pregnancy can be traced back hundreds of years[4,5]. The first proposed acknowledgement of the teratogenic effects of Prenatal Alcohol Exposure (PAE) is attributed to the biblical Book of Judges 13:3-4, from the *Old Testament*. The passage cautions Samson's mother, «[…] Behold,

you will conceive and bear a son; now do not drink wine or intoxicating drink and do not eat anything unclean, because the child will be a Nazirite[a] of God from the mother's womb until the day of his death»[4,5].

Various historical references suggest a connection between maternal alcohol consumption and congenital disabilities. One such reference is from Aristotle's Problemata, which gives another quote referenced in FASD-related knowledge descriptions: «Foolish, drunken, or hair-brain women most often bring forth children like unto themselves, morose and languid». Problemata was assembled from a variety of sources over several years and translated several times; sources that use or refer to this quote have drawn from Robert Burton's 1621 book The Anatomy of Melancholy[6] as a "primary" source and attributed as indicative of Aristotle's awareness of prenatal alcohol effects. Another reference is the Carthaginian law that prohibits alcohol consumption on the night before a couple's wedding. In 1999, Abel[7] attributed to Plato's Laws[8], also suggested that couples trying to conceive should avoid getting intoxicated. According to Plato, the embryo should be firmly, steadily, and quietly compacted in the womb and not formed by bodies dissolved by excess wine.

This law was created to prevent the conception of defective children. Although there are different interpretations of Fetal Alcohol Spectrum Disorders (FASD), these historical references provide evidence of a link between maternal alcohol consumption and congenital disabilities deriving from prenatal insult. Other researchers have also reflected on the history of FASD, supporting and contending its recognition.

Nevertheless, according to Krous[9], it would be wise to follow the Biblical instruction to abstain from wine and strong drink during pregnancy (Judges 13:7). We can ensure our well-being and safety by heeding the Biblical guidance in Judges 13:7 to refrain from consuming wine and strong drinks until more information is available. Let us make the right choice and stay on the side of caution. The story is full of references to the care one should take when drinking during pregnancy or when fertilization is supposed to occur. Despite this, the memory of this knowledge has been lost over time.

Other signals dispersed over time. Henry Fielding, referring to what happened in that time (1690-1752), which was defined as the era of the "Gin Epidemic", states: «What must become of the Infant who is conceived in Gin? with the poisonous Distillations of which it is nourished both in the Womb and at the Breast»[10].

In The Pickwick Papers, Charles Dickens introduces the character of Betsy Martin. «Betsy Martin, widow, one child, and one eye, ... but knows her mother drank bottled stout and should not wonder if that caused it... Thinks it not impossible that if she had always abstained from spirits, she might have had two eyes by this time».

Evidence of FASD also appears in paintings from the mid-19th century, when artists began to portray aspects of real life and ordinary people. With their work and sensitivity, they have grasped and represented the dysmorphological characteristics of FASD. Around 1980, Paul Lemoine, visiting the Musée d'Orsay, was able to note the presence of women who consumed alcohol in many works by Van Gogh, Toulouse Lautrec, Edgar Degas, Picasso of the blue period, Manet, Bertrand, etc. in a lonely sadness, and children and adults with the characteristics of FASD.

The FASD in Italy

In 1999, we reached out to prof. P. Kodituwakku, a neuropsychologist from the University of New Mexico, with a proposal for an epidemiological study of Fetal Alcohol Spectrum Disorders (FASD) in Italy. After several meetings with the leaders of the National Institute on Alcohol Abuse and Alcoholism (NIAAA - NIH) and a brief training course in South Africa, the NIAAA, the International Consortium for the Study of FASD (CIFASD) for the United States, the Lazio Region, and the Italian Society for the Treatment of Alcoholism and its Complications (SITAC) decided to fund an epidemiological project on the prevalence of FASD in Italy in November 2003. This study, the first epidemiological study of FASD in Europe and the second in the world, resulted in the production of scientific studies[11-16] and the dissemination of information on this syndrome, contributing to prevention efforts that we hope will become increasingly influential.

It is unfortunate to note that the recognition and diagnosis of FASD in Italy is still a challenge to date. Despite extensive research, only a handful of cases of FASD in children have been documented in Italy[17,18]. These cases depict similarities in physical and behavioral characteristics to those observed in children with FASD in the United States. It is high time that the Italian medical community takes a closer look at this issue and starts taking necessary steps toward the early diagnosis and treatment of FASD.

Training operators to identify and care for children with this syndrome is critical. An early diagnosis can prevent the most severe forms of the syndrome, which makes the chapters about diagnosis and treatment crucial. Despite the scientific evidence, difficulties still exist in understanding this syndrome. Therefore, we must focus on training operators to ensure children receive the necessary treatment.

We currently have a powerful weapon to combat FASD – widespread information and in-depth knowl-

[a] "Nazirite" comes from the Hebrew word nazir, meaning "consecrated" or "separated".

edge. Unfortunately, the interests behind alcoholic beverages often disseminate inconsistent information that contradicts scientific research, while scientific resources to combat FASD are minimal. It is crucial to implement the knowledge of professional operators to identify FASD and increase the number of authorized centers to diagnose it as early as possible to improve care for affected children.

Understanding the history and impact of FASD is essential in developing effective prevention strategies and providing adequate support for affected individuals and their families. By educating ourselves about the complexities of this diagnosis, including the need for multiple assessments such as dysmorphological, genetic, and psychological evaluations, we can work towards a future where those with FASD have access to the most suitable treatment options. Let's take action towards a brighter future where Fetal Alcohol Spectrum Disorder is no longer a prevalent issue.

These guidelines include nine publications dealing with FASD prevention, epidemiology, diagnosis and treatment.

Conflict of interests: the authors have no conflict of interests to declare.

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