DISABILITA' INTELLETTIVA ED EPILESSIA

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OUTLINE

- Caratteristiche epilessia nei soggetti con ritardo mentale
- Problemi terapeutici e diagnostici
- Quali soluzioni: parere dei medici
- Il parere dei pazienti

Caratteristica dell´epilessia (1)

- Alta incidenza - superiore alla popolazione generale (22%)
- Incidenza maggiore di epilessia in rapporto ad un quadro di maggiore MR
- Frequente esordio nell´infanzia e prosecuzione in etá adulta
- Elevata incidenza di epilessie gravi (spesso generalizzate), farmacoresistenti
- Esordio ed evoluzione spesso influenzata da eziologia (es.: Dravet s; Down)

Caratteristiche dell´epilessia (2)

Table 1 Role of IDD in epilepsy types and remission

	Remission, %	Intractable epilepsy, %
All epilepsies		
Mild IDD	49	14
Moderate IDD	32	38
Severe/profound IDD	24	55
Focal epilepsy only		
Normal intelligence	68	15
Mild IDD	57	10
Moderate IDD	28	30
Severe/profound IDD	28	44

Abbreviation: IDD = intellectual and developmental disabilities. Remission of Epilepsy in Nova Scotia study: severity of the IDD.[5]

VIEWS & REVIEWS

Delivery of epilepsy care to adults with intellectual and developmental disabilities

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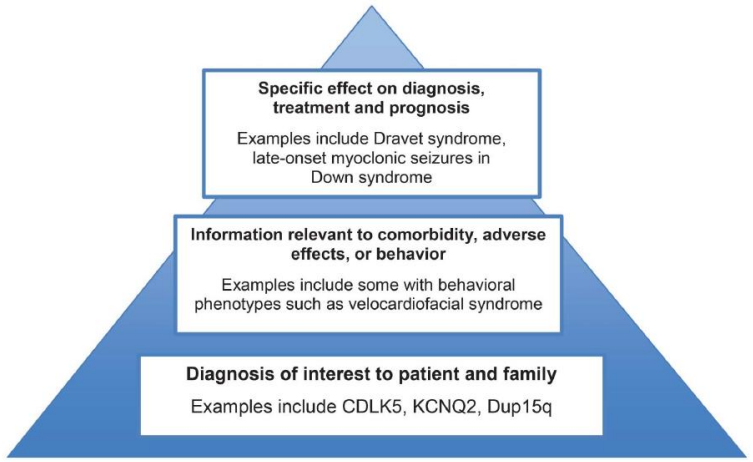
ABSTRACT

Epilepsy is common in people with intellectual and developmental disabilities (IDD). In adulthood, patients with IDD and epilepsy (IDD-E) have neurologic, psychiatric, medical, and social challenges compounded by fragmented and limited care. With increasing neurologic disability, there is a higher frequency of epilepsy, especially symptomatic generalized and treatment-resistant epilepsies. The causes of IDD-E are increasingly recognized to be genetic based on chromosomal microarray analysis to identify copy number variants, gene panels (epilepsy, autism spectrum disorder, intellectual disability), and whole-exome sequencing. A specific genetic diagnosis may guide care by pointing to comorbid disorders and best therapy. Therapy to control seizures should be individualized, with drug selection based on seizure types, epilepsy syndrome, concomitant medications, and comorbid disorders. There are limited comparative antiepileptic drug data in the IDD-E population. Vagus nerve and responsive neural stimulation therapies and resective surgery should be considered. Among the many comorbid disorders that affect patients with IDD-E, psychiatric and sleep disorders are common but often unrecognized and typically not treated. Transition from holistic and coordinated pediatric to adult care is often a vulnerable period. Communication among adult health care providers is complex but essential to ensure best care when these patients are seen in outpatient, emergency room, and inpatient settings. We propose specific recommendations for minimum care standards for people with IDD-E.

Figure 1 Hierarchical impact of genetic diagnoses on treatment of epilepsy and comorbid disorders



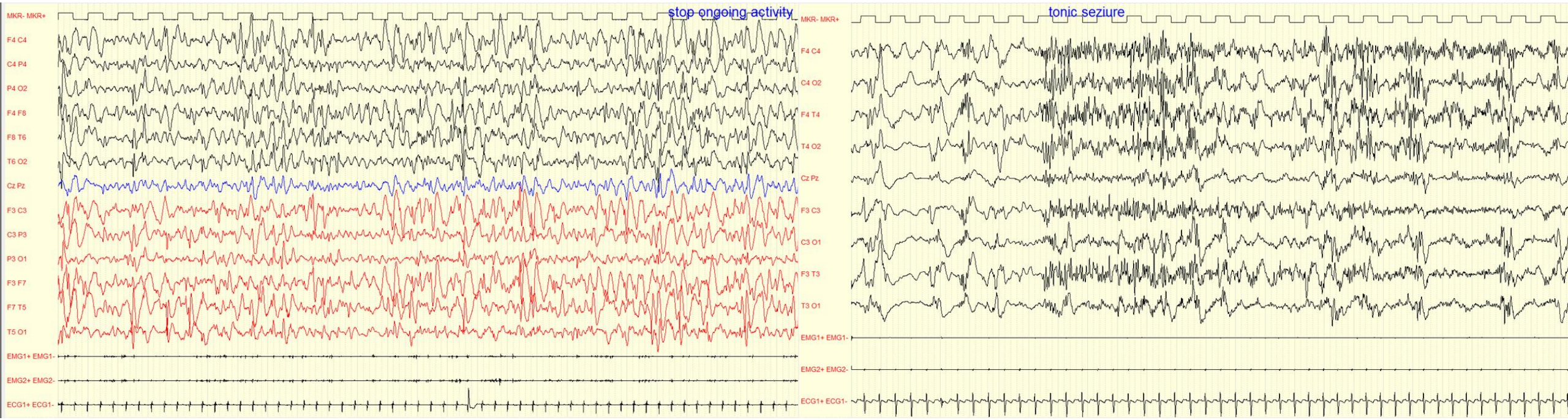
Hierarchical impact that genetic diagnoses can have on treatment of epilepsy and comorbid disorders in patients with intellectual and developmental disabilities and epilepsy. For almost all cases, genetic diagnosis provides answers and connects families to a community. In a smaller number of cases, genetic diagnosis can improve recognition of comorbid disorders and therapeutic choices.

Table 2 Examples of recurrent copy number variants associated with ID, epilepsy, and other neurodevelopmental phenotypes[50,51]

Copy number variant	Deletion or duplication	Associated phenotypes
1p36	Deletion/duplication	ID, ASD (Del), EPI (Del)
1q21.1	Deletion/duplication	ID, ASD, SCHZ, EPI
2p16.3	Deletion	ID, SCHZ, EPI
2q13	Deletion/duplication	ID, ASD, EPI (Del)
2q37	Deletion	ID, ASD, EPI
3q29	Deletion/duplication	ID (Del), SCHZ, EPI (Del)
4p16.3 (Wolf-Hirschhorn syndrome)	Deletion	ID, EPI
4q21.21-q21.22	Deletion	ID, ASD, EPI
5q35.2-q35.3 (Sotos syndrome)	Deletion	ID, EPI
7q11.22-q11.23	Deletion/duplication	ID, EPI, ASD (Dup)
9q34.3 (9q subtelomeric syndrome)	Deletion	ID, ASD, EPI
15q11.2	Deletion	ID, ASD, SCHZ, EPI
15q11-q13 (Prader-Willi/Angelman syndrome)	Duplication/deletion	ID, ASD (Dup), SCHZ (Dup), EPI
15q13.3	Deletion/duplication	ID, ASD, SCHZ (Del), EPI
16p11.2	Deletion/duplication	ID, ASD, SCHZ (Dup), EPI
16p12.1	Deletion	ID, ASD, EPI
16p13.11	Deletion/duplication	ID, SCHZ (Dup), EPI (Del)
17p12-p11.2 (Potocki-Lupski/Smith Magenis syndromes)	Deletion/duplication	ID, ASD (Dup), EPI
17p13.3-13.2 (Miller-Dieker syndrome)	Deletion	ID, ASD, EPI
17q12	Deletion/duplication	ID, ASD, SCHZ (Del), EPI
17q21.3	Deletion	ID, ASD, EPI
22q11 (Velocardiofacial/DiGeorge syndrome)	Deletion/duplication	ID, ASD (Dup), SCHZ (Del), EPI
22q11.2	Deletion	ID, ASD, EPI
22q13 (Phelan-McDermid syndrome)	Deletion	ID, ASD, EPI
Xp22.1	Deletion	ID, ASD, EPI

Abbreviations: ASD = autism spectrum disorders; EPI = epilepsy; ID = intellectual disability; SCHZ = schizophrenia.

Caratteristiche dell´epilessia (3)

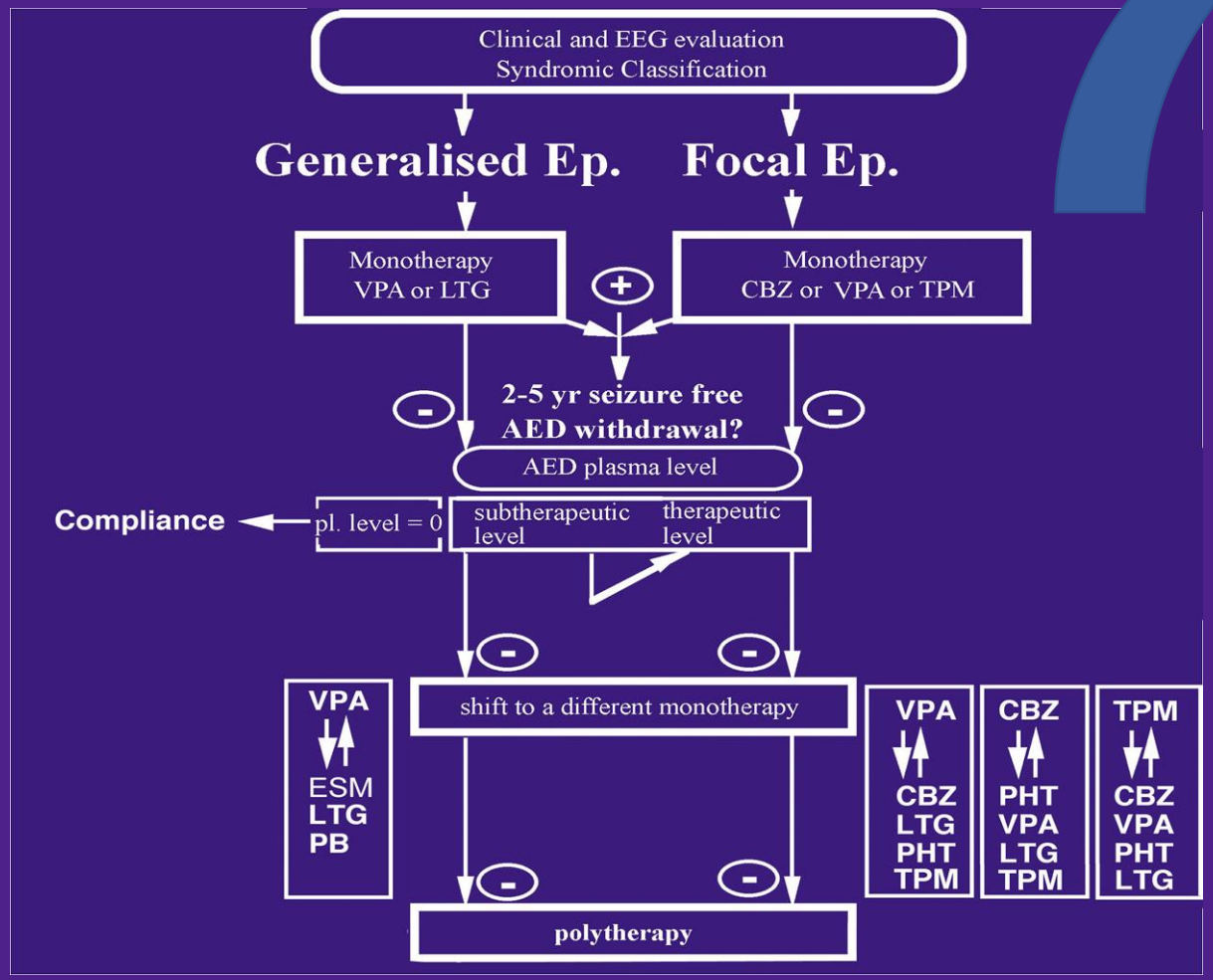


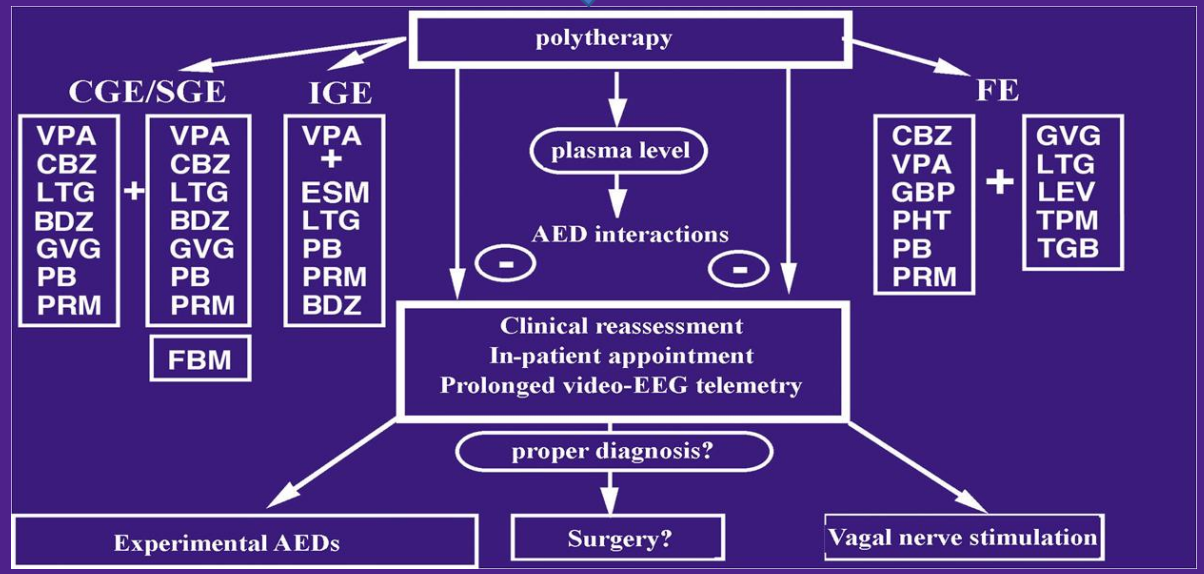
Lennox-Gastaut syndrome

Problemi diagnostici (2)

- Difficoltà nel raccogliere adeguate informazioni (paziente/caregivers)
- Difficoltà nell‘esecuzione di procedure (MRI/EEG)
- Evoluzione di quadri clinici esorditi nel bambino (lavoro anamnestico)
- Corretto inquadramento di eventi acuti (PNES vs EPI) -> video-EEG
- Expertise nella diagnosi di quadri sindromici complessi

Problemi terapeutici (1)





Problemi terapeutici (2)

- Scelta ‘ragionata’ AED –> sindromi/effetti collaterali/pregresso utilizzo
- Evitare/ridurre politerapie e attenzione alla ‘*poli-tachi-terapia’*
- Valutare sempre i ‘goal’ terapeutici con paziente/caregiver
- Non dimenticare le opzioni non farmacologiche e chirurgia
- Considerare comorbidità (psichiatriche e non)

Problemi terapeutici (3)

BUCCOLAM™

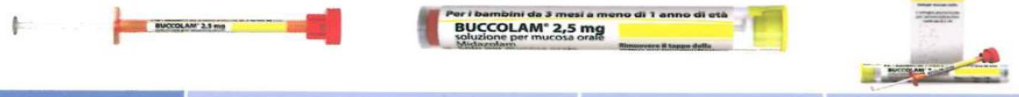
MIDAZOLAM SOLUZIONE PER MUCOSA ORALE

Benzodiazepina per mucosa orale indicata per il trattamento di crisi convulsive acute prolungate, in bambini e adolescenti (da 3 mesi a <18 anni)[1]

Posologia[1] Colori delle diverse dosi di BUCCOLAM

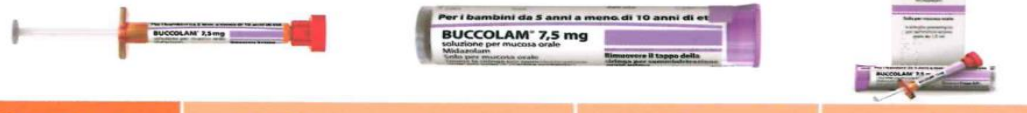
Colore dell'etichetta	Intervallo di età	Dose di midazolam	Volume della soluzione
Giallo	da 3 mesi a <1 anno[a]	2,5 mg	0,5 ml
Blu	da 1 anno a <5 anni	5 mg	1,0 ml
Viola	da 5 anni a <10 anni	7,5 mg	1,5 ml
Arancione	da 10 anni a <18 anni	10 mg	2,0 ml

BUCCOLAM 2,5 mg



BUCCOLAM 5 mg

BUCCOLAM 7,5 mg



BUCCOLAM 10 mg

[a]Per i bambini di età compresa tra 3 e 6 mesi, il trattamento deve essere eseguito in contesto ospedaliero, in cui sia possibile il monitoraggio e siano disponibili presidi per la rianimazione.

Mod. da ref. 1

MICROPAM

MICROPAM 5 mg/2,5 ml Soluzione rettale

Un contenitore monodose da 5 mg/2,5 ml contiene:

Principio attivo: Diazepam 5,0 mg

MICROPAM 10 mg/2,5 ml Soluzione rettale

Un contenitore monodose da 10 mg/2,5 ml contiene:

Principio attivo: Diazepam 10,0 mg

Eccipiente con effetti noti: alcool benzilico

Uso rettale:

bambini fino a tre anni : 5 mg

bambini oltre i 3 anni : 10 mg

adulti : 10 mg

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Intramuscular versus Intravenous Therapy for Prehospital Status Epilepticus

Robert Silbergleit, M.D., Valerie Durkalski, Ph.D., Daniel Lowenstein, M.D., Robin Conwit, M.D., Arthur Pancioli, M.D., Yuko Palesch, Ph.D., and William Barsan, M.D., for the NETT Investigators*

	LZP iv	MDZim
> 40 Kg	4 mg	10 mg
13-40 Kg	2 mg	5 mg

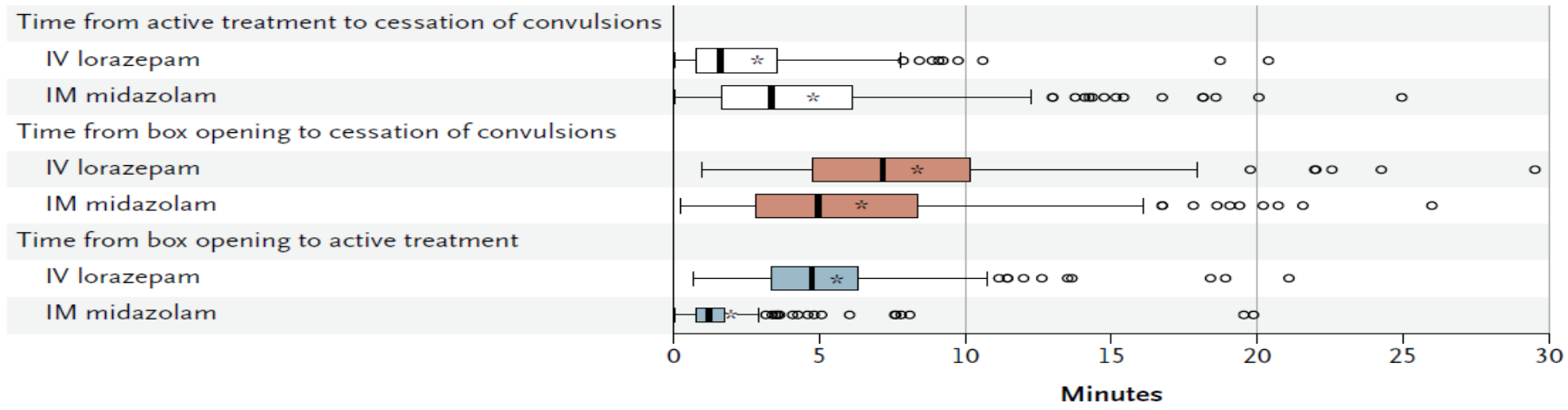

Figure 3. Intervals between Active Treatment and Cessation of Convulsions, Box Opening and Cessation of Convulsions, and Box Opening and Active Treatment.

The shorter time to IM drug administration was offset by the faster onset of action after IV drug administration, resulting in similar latency periods until convulsions were terminated. Time to IV administration includes the nominal time (about 20 seconds) needed to administer the drug by means of IM autoinjector. Asterisks indicate means, boxes interquartile ranges, bold vertical lines within boxes medians, I bars 1.5 times the interquartile range, and circles outliers.

Possibili soluzioni: parere dei medici (1)

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The provision of care to adults with an intellectual disability in the UK. A Special report from the intellectual disability UK chapter ILAE

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ABSTRACT

Purpose: This article reflects the report by the British Branch of the International League Against Epilepsy (ILAE) Working Group on services for adults with epilepsy and intellectual disability (ID). Its terms of reference was to explore the current status of aspects of the care of people with an ID and epilepsy.
Methods: Survey content was developed from key themes identified by consensus of the working group. An electronic survey was distributed via email. The sample population was the membership of the ILAE UK, Royal College of Psychiatrists (RCPsych) Faculty of ID, Epilepsy Nurses Association (ESNA), and the Association of British Neurologists (ABN). Following a six week response period the data was then collated, anonymised and distributed to the working group in order that opinion statements could be gathered.
Results: The time taken for individuals with both new-onset and established epilepsy to undergo routine investigation was commonly at least 1–3 months, far beyond recommendations made by NICE (CG20). A small minority of clinicians would not consider non-pharmacological interventions including epilepsy surgery, vagus nerve stimulation, and ketogenic diet for this population. Almost universally responders are actively involved in the assessment and management of key risk areas including risk of drowning, hospitalization, medication side effects, and sudden unexpected death in epilepsy (SUDEP).
Conclusion: This investigation identifies key themes and recommendations relating to care delivery and meeting the complex needs of people with ID and epilepsy. Adults with ID and epilepsy appear to exist in a unique, but inadequate, segment of epilepsy care delivery.
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- Questionario inviato a medici psichiatri, neurologi e infermieri
- Temi di interesse:
  - ❑ Diagnosi e trattamento
  - ❑ Rischi legati all'epilessia
  - ❑ Impatto sulla QOL
  - ❑ Modalità assistenziali
- Diagnosi: ritardo esecuzione esami, difficoltà accesso EEG, MRI
- Terapia: utilizzo sia AED, che KD, VNS e chirurgia se indicata
- Rischi legati all'epilessia: fornite informazioni esaurienti
- Impatto sulla QOL: discussa con i pazienti
- Modalità assistenziali: gestione in modo frammentario

  da figure professionali diverse (neurologi, psichiatri esperti in ritardo mentale, infermiere professionali con esperienza in epilessia)

Possibili soluzioni: parere dei medici (2)

SPECIAL REPORT

A White Paper on the medical and social needs of people with epilepsy and intellectual disability: The Task Force on Intellectual Disabilities and Epilepsy of the International League Against Epilepsy

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Epilepsia, 55(12):1902–1906, 2014
doi: 10.1111/epi.12848

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SUMMARY

This White Paper builds on the publication of the International League Against Epilepsy (ILAE) and International Bureau for Epilepsy (IBE) report "Listening for a change—medical and social needs of people with intellectual disability who have epilepsy" (Listening for a change the medical and social needs of people with epilepsy and intellectual disability, ILAE, 2013). The Paper presents an overview of the recommendations of the report, which aim to improve the health and social care of this important population of people with epilepsy worldwide. Actions in four domains are indicated: (1) the development of standards and initiatives that would enhance diagnosis, pathways to investigation, and treatment; (2) the development of guidelines for treatment, specifically best practice in the management of antiepileptic drugs including rescue medication; (3) the development of standards for primary care, multidisciplinary teamwork, and clinical consultations, with emphasis on the need to enhance communication and improve access to information; and (4) the enhancement of links among different stakeholders including medical services, educational establishments, employment services, organizations providing opportunities for social engagement, and family members. The breadth of needs of this population is a challenge to the epilepsy world, spanning all the professional groupings, care providers, and the research modalities in epilepsy.

KEY WORDS: Intellectual disability, Families, White Paper, Health care, Social care.

- Standardizzazione di percorsi diagnostici e terapie
- Sviluppo di Linee Guida per il trattamento
- Standardizzazione delle modalità assistenziali
- Creazione di un network di operatori

Il parere dei pazienti

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"Sometimes, it just stops me from doing anything": A qualitative exploration of epilepsy management in people with intellectual disabilities and their carers

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Questionario semi-strutturato a pz e caregivers:

1. Riduzione in frequenza/gravità crisi con AED
2. Impatto dell'epilessia sulla QOL per tutta la vita
3. Percezione della difficoltà a convivere con l'epilessia ed a trovare una terapia adeguata
4. Percezione di un adeguato livello di assistenza
5. Mancanza di adeguate informazioni scritte sull'epilessia

Conclusioni

- Epilessia spesso farmacoresistente e grave
- Accesso a diagnostica e a terapia non sempre soddisfacente
- Necessità per gli operatori di agire in Team work
- Utilità di networks di professionisti con competenze adeguate
- Importanza di progetti come il DAMA (Disabled Advanced Medical Assistance)


Grazie per l’attenzione