

# Epilepsy in Intellectual Disabilities



## Dr Howard Ring

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### Topics addressed in the article

- What is Intellectual Disability (ID) ?
- A brief account of the nature of epilepsy in people with ID.
- Issues around the diagnosis of epilepsy in people with ID.
- Education and communication in the management of epilepsy in people with ID
- Neuropsychiatric comorbidities of epilepsy in people with ID.
- The evidence base informing management of epilepsy in people with ID.

### What is Intellectual Disability?

To have an intellectual (learning) disability (ID) is to have a developmental disorder characterised either by never having been able to acquire the educational and functional skills expected for your age, or, early in life suffering a neurological insult that arrested your development such that you could not go on to develop the expected level of functioning. Whatever the cause, those considered as having an ID manifest significantly limited abilities across a wide range of everyday functions including cognitive, language, motor and social activities. In the UK the diagnosis of an ID also requires IQ to be 70 or less. An estimated 828,000 adults in England have an ID and amongst this population epilepsy is common, being the most frequent medical illness experienced.<sup>1</sup>

### The nature of epilepsy in people with ID

Across the population of those with ID as a whole, a prevalence of epilepsy of 26% has been reported though this average figure obscures the fact that prevalence of epilepsy increases in line with increasing severity of ID. Amongst those with mild to moderate ID lifetime epilepsy prevalence has been reported at between 6 and 15%. In those with severe ID epilepsy occurs in around 25% whilst in those with profound ID (IQ<20), in whom it has been estimated that there will in the UK be an average annual increase in number of 1.8%, epilepsy is reported in more than 50%. In some specific ID syndromes particularly high rates of epilepsy are reported; for instance in Rett and Angelman syndromes prevalence rates for epilepsy of 80% or more are reported.

Not only is epilepsy more common in those with ID than in the rest of the population: it tends to have a worse prognosis, with lower rates of seizure freedom<sup>4</sup> and high rates of multiple antiepileptic drug use,<sup>5</sup> incurring more side-effects<sup>6</sup> and higher treatment costs.<sup>7</sup> Adults with ID and epilepsy have high rates of morbidity<sup>8</sup>

and mortality,<sup>9</sup> including sudden unexplained death in epilepsy (SUDEP).<sup>10</sup> Indeed, the Standardised Mortality Ratio (SMR) for SUDEP in adults with intellectual disability and epilepsy is in excess of 30.<sup>10</sup>

There appear to be multiple aetiologies underlying the association between epilepsy and ID and this is currently a very active area of research that in the future may suggest novel treatment approaches. Aetiological processes include not only effects of well-described genetic anomalies such as those leading to Rett and Angelman syndromes, but in an as yet undetermined proportion of people with ID and epilepsy the effects of an unknown number of rare but clinically significant submicroscopic copy-number variants (CNVs).<sup>2</sup> There is also evidence from experimental research to suggest that changes associated with epileptogenesis and seizures in early post-natal life may have effects on developmental processes in the brain including disruption of synaptic plasticity, dendritic development and ion channel maturation that may lead to later impairment in cognitive development.<sup>3</sup>

### Diagnosis of epilepsy in people with ID

The diagnosis of epilepsy in people with ID may be complicated by a range of issues including; conflicting eye witness accounts of possible seizure events together with the difficulty that the patient themselves is likely to have in providing a history; the presence, particularly in those with more severe or profound ID, of stereotyped movements or mannerisms that may be mistaken for seizure-related movements; other paroxysmal disturbances of behaviour, for instance related to pain or frustration; and, potentially further compounding the challenges in clarifying the diagnosis; the difficulty that some people with ID may have in tolerating investigations such as EEG and MRI. Evidence suggests that in people with ID there are significant rates both of misdiagnosis of non-epileptic seizures as epilepsy, in up to around a third of cases, and failure to diagnose or to treat episodes that are epileptic in nature.<sup>11</sup> Hence it is important when managing refractory epilepsy in people with ID that the diagnosis is carefully reviewed and at the same time episodes considered to be behavioural in nature are re-considered to check that an epilepsy diagnosis is not being missed.

### The role of education and communication in management of epilepsy in people with ID

Unlike most of the population without an ID, many of those who do have ID also rely for

some or all of their day-to-day support on family or on paid care workers. Hence not only can poorly controlled epilepsy impact negatively on quality of life of people with ID and epilepsy – it may also increase demands on families and others who provide support and care. Clinical and research evidence demonstrates that in order to deliver epilepsy management well to people with ID, it is important to appropriately involve this wider circle of individuals. This involvement should include good communication and, importantly, training by appropriate healthcare professionals of the people that support those with ID and epilepsy in the community. This is a critical element of care and one that often does not have a counterpart in epilepsy management, at least of adults, in the rest of the population. Another important consequence of this reliance that adults with ID and epilepsy have on family or paid supporters is that a key element in the therapeutic relationship that clinicians should focus on is their relationship with these supporters. Research has demonstrated that this is important in contributing to the transmission of relevant observations to the clinicians and to the potential uptake of and compliance with suggested antiepileptic therapeutic interventions offered to patients.<sup>12</sup>

### Neuropsychiatric comorbidities of epilepsy in people with ID

There is evidence that some psychiatric comorbidities are more common in those with epilepsy who also have an ID and amongst these the conjunction of autism and epilepsy is well recognised, with autism occurring in up to 30% of people with epilepsy,<sup>13</sup> most often in those who also have an ID. In a recent study investigating the details of this relationship it was noted that the frequency of cases positive for epilepsy amongst a group of people with autism was highest in those whose autism was associated with an early age of diagnosis and high rates of repetitive object use and unusual sensory interests.<sup>14</sup> With respect to seizure semiology in autism, all seizure types are seen in people with autism,<sup>15</sup> with those most commonly observed being focal seizures with altered awareness, atypical absence and generalised tonic-clonic seizures. There

is no evidence that antiepileptic drug efficacy differs between those with and without autism in addition to epilepsy.

### The evidence base informing management of epilepsy in people with ID

Evidence that epilepsy in adults with ID may not be optimally managed comes from a report by the Learning Disability Observatory into Ambulatory care sensitive conditions (ACSCs) (defined as conditions which, given 'effective management' at the primary care level, should not normally result in an admission to hospital) in people with ID. That report<sup>16</sup> noted that convulsions and epilepsy were the most frequent cause of what were considered as potentially avoidable hospital admissions in people with ID, accounting for approximately 6000 admissions a year, equivalent to 40% of all emergency admissions for ACSCs in adults with ID.

Despite the frequency and potential severity of epilepsy in people with ID, many of the clinical trials that have investigated antiepileptic drug use in epilepsy management have excluded those with ID. Hence there is limited research evidence to inform clinical epilepsy management strategies among people with ID beyond that which can be extrapolated from the rest of the epilepsy population. However, a systematic review published in 2009 of the available evidence concluded that AEDs effective in the general epilepsy population are also effective in refractory epilepsy in people with ID, though conclusions on relative efficacy between medications could not be drawn.<sup>17</sup>

In order to inform treatment choices in the absence of a wider evidence base, pragmatic consensus clinical guidelines have been developed,<sup>18</sup> which supplement existing guidelines for epilepsy care in the non-intellectual disability population. They draw attention to a range of issues including; the associated communication difficulties experienced by people with ID and the possible consequences of these for detecting antiepileptic drug treatment-related adverse effects; the fact that presence of ID is not necessarily a contraindication to neurosurgery for epilepsy; and the relatively high rates of other comorbidities experienced by people with ID and epilepsy. ♦

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