Llywodraeth Cymru
Welsh Government

GUIDANCE, DOCUMENT

Early developmental impairment or intellectual disability (WHC/2023/03)

Guidelines for paediatricians in supporting clinical decisions about early developmental concerns.

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Welsh Health Circular

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Enclosures:

Guideline for the investigation of moderate or severe early developmental impairment and intellectual disability.

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who assess children and young people referred because of early developmental concerns. It establishes a baseline for Wales-wide practice to ensure equitable access to timely diagnosis wherever a child lives in Wales.

2. The All Wales Community Child Health Network, led in this work by Dr Bethan McMinn and supported by the Wales Rare Diseases Implementation Group, has produced this guidance document, which is informed by the published evidence base and professional consensus. This document supersedes any previous guidance on investigation of early developmental impairment or intellectual disability (EDI/ID).
3. The guideline aims to outline a consistent Wales-wide approach to the identification, clinical assessment and investigation of children and young people who have moderate or severe EDI/ID.
4. The guideline offers a standardised approach to diagnostic (including genomic) testing, to reduce the diagnostic odyssey, promote diagnostic yield, efficiency and equity of access, identify any treatable causes and also to identify any clinical features or ‘red flags’ which would indicate more targeted or specialist testing is indicated.
5. The guideline sets out in Table 1 the recommended first line investigations for children with isolated moderate or severe EDI/ID. The inclusion of more recent genetic testing techniques has increased diagnostic yield, but please note the need for specific consent for this testing. Bilingual parent information leaflets are included with the guideline.
6. Second line investigations might be indicated if uncertainty remains. These are recommended at clinical discretion, which might follow repeat assessment or a “watch and wait” approach. Additional specialist advice may be sought from medical genetics.
7. The guideline appendices contain a summary flowchart, a table of clinical features additional to EDI/ID that might prompt targeted testing, and a table to summarise results.
8. The guideline should be implemented as soon as possible to promote best quality services for children with EDI/ID. Clinical networks should audit diagnostic timeliness and patient satisfaction.

Link to the '**Guideline for the investigation of moderate or severe early developmental Impairment and intellectual disability**.