Global Developmental Delay in Pre School Children

Guidelines for Investigation

Global developmental delay is defined as significant delay in two or more developmental domains.

Investigation should be considered only after a thorough history and examination have been performed.

These guidelines are not intended for isolated speech and language or motor problems, or for children with autism.

- If diagnosis not apparent after history and examination, proceed as follows:

  **First Line**
  - Chromosomes
  - Fragile X
  - U & E
  - Creatine kinase
  - Lead
  - Thyroid function tests
  - Urate
  - Full blood count
  - Ferritin
  - Biotinidase

  **Second Line**

  - **Metabolic**
    - Family history
    - Consanguinity
    - Regression
    - Organomegaly
    - Course features

  - **Neuroimaging**
    - Abnormal head size
    - Seizures
    - Focal neurology

  - **EEG**
    - Speech regression
    - Seizures
    - Neurodegenerative disorder

  - **Genetics**
    - Dysmorphism
    - Abnormal growth
    - Sensory impairment
    - Odd behaviour
    - Family history

  - **Blood**
    - Lactate
    - Amino acids
    - Ammonia
    - VLCFA
    - Carnitine
    - Homocysteine
    - Disialotransferrin

  - **Urine**
    - Organic acids
    - Oxalate
    - Gags
    - Oligosaccharides

  - **MRI**
    - CT (bones, calcification)

  Consider 24 hr EEG