

Investigation of global developmental delay

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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
Coarse features



Blood

Lactate
Amino acids
Ammonia
VLCFA
Carnitine
Homocysteine
Disialotransferrin

Urine

Organic acids
Orotate
Gags
Oligosaccharides

Neuroimaging

Abnormal head size
Seizures
Focal neurology



MRI

CT (bones, calcification)

EEG

Speech regression
Seizures
Neurodegenerative disorder



Consider 24 hr EEG

Genetics

Dysmorphism
Abnormal growth
Sensory impairment
Odd behaviour
Family history