Clinical Guideline

INVESTIGATIONS GUIDE FOR CHILDREN WITH DEVELOPMENTAL DELAY

CARE PATHWAY	Developmental Delay Care Pathway
FOR STAFF	Community Paediatrics
SETTING	Outpatients
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GUIDANCE

This guideline for the investigation of developmental delay is for use alongside the CCHP Developmental Delay Care Pathway. This tool is primarily to support clinicians in planning to investigate causes of Developmental Delay in a child or young person.

Definitions

Developmental delay is defined as significant delay (more than two standard deviations below the mean) in one or more of the following developmental domains:

- Gross motor
- Vision & Fine motor
- Hearing, Speech & Language
- Social, Emotional & Behavioural

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

Initial Assessment

- Full history and examination including neonatal period, consanguinity, episodes of hypoglycaemia
- Developmental assessment
- Hearing and vision assessments
- Woods lamp
- Growth parameters including head circumference

Investigations

These are guidelines for sequential investigations and must be considered in the context of an individual's history and examination. Non-sequential investigations could be considered in the interests of opportunistic testing.

First line:

Genetic blood tests:	**Standard blood tests:**	**If indicated*:**	**Urine:**
• CGH Microarray • Karyotype *	• TFTs • CK • Urate, chloride • FBC & Ferritin • U&E • LFT • Calcium	• Biotinidase* • Lead* • Acyl Carnitine profile* • TORCH Screen*	• Glycosaminoclycans (MPS screen)*

*See Table (Appendix 1) for further information re indication and cost.

Second line:

- **Metabolic;** If family history, regression, consanguinity, abnormal head size, organomegaly, coarse features, seizure, abnormal neurology.

  Blood tests:
  Lactate
  Ammonia
  Amino acids
  Homocysteine
  Biotinidase, FBC, U&E, LFT, Calcium, AcylCarnitine profile if not already done
  VLCFA
  White cell enzymes

  Urine:
  Sialyloligosaccharides
  Organic acids

- **Neuroimaging**

  MRI; If abnormal neurology, abnormal head size, seizures, and vision problems.
  CT; Only where cerebral calcification is suspected e.g. in perinatal infection or to look for abnormality of skull bones.

- **Neurophysiology**

  EEG; If abnormal neurology, seizures, regression of language, neurodegenerative disorder.

- **Genetics**

  Fragile X, Test - for Specific genetic disorder, Methylation/ UPD/ Mutation studies
  Referral to clinical genetics*

Third line:

Potential third line blood tests (consider neurology, metabolic and genetic referrals by this stage)

- **Metabolic**

  Iso-electric focussed Transferrin
  7-dehydrocholesterol
  Blood gas or chloride to calculate anion gap if not done already
  Paired plasma creatinine and urine guanidinoacetate

APPENDIX

1) Investigations for Developmental Delay; Cost and Interpretation
2) Investigations for Developmental Delay Results Table
3) Investigations for Developmental Delay Flow Chart

OTHER RELATED DOCUMENTS Nil

REFERENCES

References:
1.McDonald, A., Rennie, J., Galloway, P. & Mc William, R. (2006) Investigation of Global Developmental Delay, Arch Dis in Child, 91, 701-705
2. Walters, V. A. (2010) Developmental Delay Causes and Investigation. Advances in Clinical Neuroscience and Rehabilitation, 10-2,32-34.
3. Sharma A (2011) Developmental examination: birth to 5 years, Arch Dis Child Educ Pract Ed. 2011 Oct;96(5):162-75. doi: 10.1136/adc.2009.175901.
4. Srour M, Shevell M (2013) Genetics and the investigation of developmental delay/intellectual disability, Arch Dis Child doi:10.1136/archdischild-2013-304063

SAFETY Nil known

QUERIES Dr Latha Chandramouli

AUDIT TOOL TBD