Paediatric Genomics Update: 2020 Medicare funded genomic testing in paediatrics

Introduction
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By the end of this session, paediatricians will be able to:

1. Recognise when genomic testing would be suitable for a patient
2. Consider genomic testing:-
   a. Which test to order
   b. Obtain consent
   c. Organise genomic testing on a patient
3. Explain a genomic test result
4. Know when to contact Genetics Services for assistance

Paediatric Genomics Update

Learning outcomes:
Genomic testing in paediatrics will be Medicare funded from May 1st 2020

- Genomic testing = ALL Genes and DNA
- Genetics = single gene(s)
- As requested by a clinical geneticist or paediatrician, in consultation with a clinical geneticist
- For patients ≤ 10 years of age, with:
  - Dysmorphic features and 1 or more major structural congenital anomalies or
  - Intellectual disability (ID) or global developmental delay of at least moderate severity
- Final details of Medicare item released on 1 May 2020.

Genomic Testing Pathway

Any child ≤ 10 with:
- Dysmorphia and ≥ 1 Congenital structural anomalies or
- Moderate Intellectual Disability

Chromosome Microarray / Fragile X/Urine Metabolic Screen

Consider Genomic testing:
Which genomic test? / Obtain Genomic Consent / How to organise the test?

Explain possible genomic results

- Genetic cause identified
  - Genetic diagnosis, causative variant found
  - May need additional clinical correlation and counselling
- Variant of uncertain significance
- Incidental finding
  - May need additional clinical correlation and counselling
- Negative result
  - May benefit from reanalysis in 2 years

Refer/discuss with genetics at any stage
If concerns regarding pregnancy planning, consent, results interpretation
www.genetics.edu.au
Take Home messages

1. Recognise when genomic testing would be suitable for a patient
   • Any child <10 years of age with: Dysmorphism and ≥1 congenital structural anomalies OR moderate Intellectual Disability/Global Developmental delay
   • After normal CMA, Fragile X & UMS
   • Ensure it’s appropriate for family

2. Obtain genomic consent and order genomic testing on a patient
   • Trio whole exome sequencing best especially for ID/neurodevelopmental disorders
   • Consent for patient (singleton) OR patient and both parents (trio)
   • Send 2-10mls EDTA to local lab for “DNA extraction and storage”, “WES” after checking lab requirements
   • Liaise with local genetics service regarding lab and send with exome request:
     - consent for patient or x 3 if trio
     - Phenotypic/clinical data (Medicare requirements)

3. Explain a genomic test result
   • Genetic cause identified
   • VOUS
   • Incidental findings
   • Negative (can re-analyse in 2 years)

4. Know when to contact Genetics Services
   • ANYTIME
   • Details of Genetics Services: www.genetics.edu.au

Refer or discuss with Genetic Services at any time or stage

Find information at:
The Centre for Genetics Education NSW Health
www.genetics.edu.au
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