

# Diagnosis - the forgotten step?

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# Developmental Delay

Screening



Diagnosis



Treatment / Intervention

Screening is a strategy used in a large population of apparently well people to detect individuals who *may* have an illness / developmental delay

The identification of the nature and cause of an illness / developmental delay

# Developmental Delay in Tassie

Screening



Diagnosis



Treatment / Intervention

- CHAPS nurses
- General Practice nurses (4 yr old checks)
- GPs
- Allied Health

- Paediatricians
- GPs
- Allied health (in their area of expertise - nature more than cause)

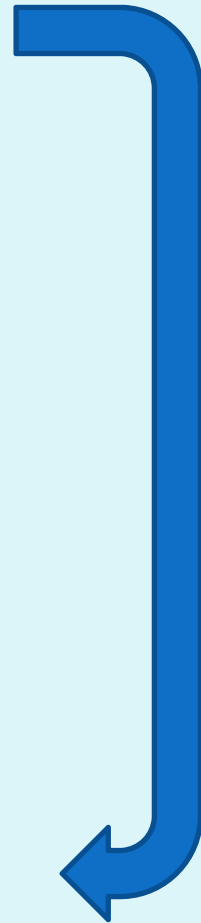
- Public and Private intervention providers
- GP s and Paediatricians

# Developmental Delay in Tassie

Screening

Diagnosis

Treatment / Intervention



# Diagnosis of Developmental Delay

Diagnosis attempts to provide not just a definitive description of the problem  
(exactly how delayed is the child and in what areas)

But also an answer to the question WHY is the child delayed? What is the CAUSE of the problem?

# Why is Diagnosis Important?

- Appropriate treatment / interventions
- Prognostic information
- Diagnosis-specific supports / funding
- Recurrence risk information
- Epidemiological data
- Provides 'an answer' / limits unnecessary testing / remove guilt

# How does a Paediatrician make a Diagnosis?

- Complete History
- Examination
- Investigations

## First Line Tests

(reasonable evidence to support routine use)

Hearing and Vision

FBE

Iron studies

CK

TFTs

Microarray

Fragile X test

Urine Metabolic Screen

## Second Line Tests

(consider on a case by case basis)

MRI brain

EEG / sleep EEG

Metabolic testing

Lead levels

transferrin isoforms

[there are many more...]

# How does a Paediatrician think about causation?

## PRENATAL

Chromosomal - aneuploidy , deletions / duplications

Single gene disorders - Fragile X , Duchenne, Rett, Prader Willi, Angelman, Tuberous sclerosis

Complex genetic disorders - Inherited MR , X-linked MR, ASD

Teratogens - Medications, Recreational Drugs / Alcohol

Maternal Infections – TORCH

Maternal Thyroid and Iodine status

Cerebral Malformations

## PERINATAL

Prematurity

Birth Asphyxia

Infection

Trauma

## POSTNATAL

Infection

Trauma

Tumour

Stroke

Seizure

Metabolic Disease

Psychosocial deprivation

# How often can a cause be found?

Depends on the sample and level of testing  
Few studies in which same battery of tests given  
Variable reports of diagnostic yield between 10 – 81%

**BUT**

In unselected Global Dev Delay , most studies report rates around...

**30%**

[Srouf et al. Pediatrics Vol 118, No 1, July 2006]

## My Message...

All children with Developmental Delay should be seen by a Paediatrician to ensure a thorough diagnostic assessment is undertaken.

# Some Examples...

RB

5 yr boy

GDD

FSIQ 56 (high achieving family)

Clinodactyly

Maternal guilt about alcohol in pregnancy +++

Microdeletion @ 20q13  
of 1.3 Mb [50 genes]

Relief of maternal guilt

Provision of 'an answer'

Further testing avoided

Prognostic information

# Some Examples...

MA

4 yr 6 mth girl

Language delay

Social difficulties

FSIQ 75

VSD and ASD

Fragile X Pre-mutation

Provision of 'an answer'

Prognostic information – ASD / learning / premature ovarian failure in 20%

Recurrence risk – cascade testing

# Some Examples...

AH

7 year old boy

Facial Dysmorphism – no syndrome diagnosis

Severe language delay (vocalised but few words rarely used)

WISC at 5 yrs : performance IQ 98, language sections not able to complete

Speech path several years: PECS and Makaton used well

Father reported he often pulled at right ear, TV up loud

Left ear: Middle ear fluid  
Right ear: Cholesteatoma  
Audiology: severe hearing deficit

ENT surgery

Hearing Aids bilaterally



Thank You!