

## Basics to the Approach of Developmental Delay

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### Introduction

Developmental disability is estimated to occur in 5-10% of the population with enormous psychological, emotional, and economic impact on the affected individuals and society. Studies have shown that developmentally delayed children who are recognized at an early age receive more developmental optimization and greater gains than those who are identified later in life. Early recognition of children with developmental problems is therefore important.

A child who does not reach his or her developmental milestones at the expected age is considered developmentally delayed. [See table summarizing developmental milestones](#). There are several disabilities in the classification of developmental delay:

| Disability   | Description  |
|--|--|
| Gross motor delay  | Significant delay in fine or gross motor skills with no impairment in other developmental areas  |
| Developmental language disorders   | Significant delay in receptive and/or expressive language skills with no delay in other developmental domains  |
| Global developmental delay   | Significant delay in two or more developmental streams as measured by appropriate standardized screening tests. This term is reserved for children less than 5 years of age                    |
| Cerebral palsy   | Early-onset non-progressive motor impairment with associated abnormalities in muscle tone  |
| Visual sensory impairment  | An optically or medically diagnosable condition in the eye(s) or visual system that affects the development and normal use of vision, ranging from slight to complete blindness                |
| Hearing sensory impairment   | A reduction in the ability to hear sound, ranging from slight to complete deafness   |
| Learning disabilities  | Significantly lowered individual achievement than predicted by intellectual ability as measured by standardized psycho-educational tests assessing reading, mathematics, or written expression |
| Pervasive developmental delay (PPD) / Autism   | Impairments in social skills, communication skills and restrictive / repetitive patterns of behavior   |
| Pervasive developmental disorders not otherwise specified / Autism Spectrum Disorder | Similar to PDD but not enough symptoms to warrant a PDD diagnosis  |

### An Approach to a Child with Developmental Delay

A child's development is a dynamic process, and assessment at any point in time is merely a snap shot of the bigger picture and should be interpreted in the context of the child's history from conception to the present. While a child may appear to have normal development for the first twelve months of life, a deviation in the course of the child's development in subsequent years is indicative of an underlying disability. It is important to keep this in mind as you assess a child, and to keep reassessing children in subsequent office visits.

Developmental assessment involves three aspects: screening, surveillance, and definitive diagnostic assessment.

- 1) *Developmental screening* is identifying children who may need more comprehensive evaluation. It is a brief assessment procedure designed to identify children who should receive more intensive diagnosis or assessment. This is accomplished in the pediatrician's office through thorough history taking +/- the use of screening tools such as the Denver or Bayley Scales of infant development.
- 2) *Developmental surveillance* is a continuous process whereby the child is followed over time to pick up on subtle deficiencies in the child's developmental trajectory. The components of developmental surveillance include eliciting and attending to parental concerns, obtaining a relevant developmental history, observing the child's development in the office and referring for further assessment of development by other relevant professionals such as OT/PT for motor developmental concerns or hearing tests for concerns with language acquisition.
- 3) *Diagnostic assessment* is performed on a child who has been identified as having a potential problem. This step requires extensive involvement of various team players such as a psychologist, educator, social worker, developmental pediatrician, geneticist, and/or other medical professionals.

### **History**

To perform a developmental assessment, a detailed history from conception to the present is required to assess developmental level. Knowing the appropriate milestones ([Table](#)) is key to this assessment. Any signs of developmental regression should be regarded as a medical emergency and an urgent medical workup is indicated.

An underlying etiology for developmental delay should be sought through attention to the following clues on history:

- 1) Prenatal history
  - a. Complications
  - b. Prenatal diagnoses made (eg. Down Syndrome)
  - c. Infections (eg. TORCH)
  - d. Exposures (eg. Fetal Alcohol Syndrome)
- 2) Obstetrical history
  - a. Complications
  - b. APGAR scores
  - c. Infections (eg. Group B Strep)
  - d. Seizures
  - e. Hearing test performed
  - f. Newborn screening performed
- 3) Past medical history and medications
  - a. Ototoxic antibiotics eg. Gentamicin
  - b. Frequent ear infections may lead to effusions affecting hearing
- 4) Behavior since birth
  - a. Behavioral disturbances – aggression, self injury, defiance, inattention, anxiety, depression, sleep disturbances, stereotypic behaviors, poor social skills, hyperactivity, difficult temperaments
- 5) Family history
  - a. Relatives with developmental delay, genetic abnormalities, syndromes
  - b. Consanguinity
- 6) Social History
  - a. Evidence of neglect or abuse which may have a negative influence on development.

- b. Primary languages. ESL children may have relative delay in English language acquisition.
- c. In children with a previously identified delay it is important to assess the resources already accessed to support the family such as personal tutors in the educational system, OT/PT for speech and language therapy, etc. The "Infant development program" is a regional resource supplying support to children until age 3. The "At home" program is a federal incentive to provide financial support and respite care to families with an affected child.

### **Screening Tools**

There are various screening instruments used for assessing developmental. The *Denver II* assesses gross motor, fine motor, adaptive and social skills. It is designed for children between the ages of 0 and 6. Similar tools are the "Ages and Stages" questionnaires and the Bayley infant development scales.

### **Physical Examination**

A thorough physical examination is important in the assessment of a developmentally delayed child. Characteristic findings on physical exam may provide clues as to the cause of the developmental delay. Some clinical signs and their corresponding clinical significance are listed below:

#### Growth Parameters:

- Microcephaly: eg in Rett's Disorder
- Macrocephaly: eg in hydrocephalus
- Short stature: Turner syndrome, Williams syndrome
- Obesity: Prader-Willi syndrome, Beckwith-Wiedemann syndrome

#### Head and Neck

- Flat occiput: Down syndrome, Zellweger syndrome
- Prominent occiput: trisomy 18
- Craniosynostosis: Crouzon syndrome, Pfeiffer syndrome
- Midface hypoplasia: Fetal Alcohol Syndrome (FAS), Down syndrome
- Prominent nose and chin: Fragile X syndrome
- Round facies: Prader-Willi syndrome
- Triangular facies: Turner syndrome
- Hypertelorism: Fetal hydantoin syndrome
- Hypotelorism: maternal PKU effect
- Brushfield spots: Down syndrome
- Prominent eyes: Beckwith-Wiedemann syndrome
- Lisch nodules: neurofibromatosis
- Large pinna: Fragile X syndrome
- Malformed pinna: Treacher Collins syndrome, CHARGE association
- Broad nasal bridge: Fragile X syndrome
- Low nasal bridge: Down syndrome
- Long philtrum: FAS
- Cleft lip and palate: may either be isolated or part of a syndrome
- Micrognathia: Robin sequence
- Macroglossia: Beckwith-Wiedemann syndrome
- Abnormal hair whorls: Down syndrome
- Webbed neck: Turner syndrome

#### Genitourinary

- Macroorchidism: Fragile X syndrome

- Hypogonadism: Prader-Willi syndrome

#### Extremities

- Small hands: Prader-Willi syndrome
- Clinodactyly: trisomies including Down syndrome
- Transverse palmar crease: Down syndrome

#### Skin

- Nail hypoplasia or dysplasia: FAS
- Facial port wine hemangioma: Sturge-Weber syndrome
- Café au lait spots: Neurofibromatosis
- Ashleaf spots: Tuberous Sclerosis

#### Neurological Exam

- Cranial nerves
- Specific vision tests
  - red reflex, normal fundi, response to visual stimuli, field of vision
- Specific auditory tests
  - response to auditory stimuli
- Receptive or expressive language delay
- Abnormal speech (eg. articulation)
- Persistently present Babinski response (older than 2 years of age)
- Hyper- or Hypotonia
- Sensory
- Motor strength
- Gait
- Deep tendon reflexes
- Primitive reflexes – Moro, Gallant
- Postural reflexes – propping response

#### Investigations

- 1) Genetics:
  - a. Karyotyping to assess for chromosomal abnormalities
  - b. FISH analysis to assess for microdeletions
  - c. Many of these investigations will be performed through specialist referral. Medical Genetics consultation should be done at this time.
- 2) Endocrinology:
  - a. TSH, free T4
  - b. Referral to endocrinology should be considered.
- 3) Metabolic:
  - a. Metabolic screening – glucose, electrolytes, serum lactate, ammonia, liver function tests, pyruvate, albumin, triglycerides, uric acid, serum quantitative amino acids, urine organic acids, acylcarnitines, creatine phosphokinase (if suspecting myopathy)
  - b. Referral to metabolic diseases should be considered.
- 4) Neurology:
  - a. EEG
  - b. Head CT
  - c. Referral to Neurology if any of these tests are considered.