

Diagnostic Aspects of Early Intervention Services

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Case I: Petra



- 9 months old
- Not sitting independently

Complete Medical History

- Was the birth normal?
- Any significant illnesses or injuries?
- Loss of developmental skills?
- Other family members with problems?

Medical Examination

- General examination
- Neurodevelopmental examinations
 - Muscle function
 - Reflexes
 - Gross motor level
 - Movement

Muscle function

- Hypertonia (stiff)



- Hypotonia (floppy)



Reflexes



- Tendon reflexes
- Primitive reflexes
- Equilibrium responses



Gross Motor Level



- Motor skill age
- Motor quotient
 - Motor skill age \div chronological age
 - E.g. 6 months \div 12 months = 50%
 - G.M. quotient $<$ 50% suggests cerebral palsy
- **Correct for prematurity**

Movement

- Stability
- Symmetry
- Isolation of movements
- Atypical
- Abnormal



Further Tests

- Neuroimaging
 - CT scan or Magnetic Resonance imaging
- Genetic
 - Chromosomes
 - Specific genes
- Metabolic
 - Amino and organic acids
 - Metabolic screens
- Further testing
 - Cognitive, motor, speech/language

Case 2: Marcus

- 2 ½ years old
- “Not talking”



Possible Causes

- Developmental delay (mental retardation)
- Autism spectrum disorder
- Specific language disorder (e.g. verbal apraxia)
- Hearing impairment
- Severe environmental deprivation
- Elective mutism
- Normal variation

Diagnostic Evaluation

- History and physical examination
- Hearing test
- Full cognitive, language, and behavioral evaluations
- Specific evaluations such as the Autism Diagnostic Observation Schedule (ADOS)
- Chromosome analysis and Fragile X DNA

Discussion with Parents

- Full disclosure of all information
- Provide written material
- Opportunity for follow-up discussion
- Balance between honesty and hopefulness
- Referral for family support services

