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Atypical Cerebral Palsy: Edmonton Experience

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Abstract: ***Cerebral Palsy is a chronic condition affecting body, limb movement and the control of muscle tone and coordination. It is caused by damage to one or more specific areas of the developing brain. The brain damage is not progressive. The diagnosis is based on the history and clinical examination. A crucial point in the history is that the delay is not progressive. In this study, a cohort data of patients previously diagnosed with cerebral palsy was investigated. We found that the presence of atypical features in the clinical presentation, laboratory, and neuroimaging profiles had guided to different diagnoses other than cerebral palsy.***

Keywords: Cerebral Palsy, Dyskinesia, Spasticity, Disability

1. Introduction

Cerebral Palsy (CP) is a chronic condition affecting body, limb movement and the control of muscle tone and coordination. It is caused by damage to one or more specific areas of the developing brain. The brain damage is not progressive; however, the characteristics of disabilities resulting from brain damage often change over time. The diagnosis of CP is based on history and clinical examination. A crucial point in the history is that the delay is not progressive. No laboratory tests are required to make the diagnosis [1].

2. Literature Survey

International Cerebral Palsy registries have shown a consistent prevalence globally: 2.0-3.5 in the Unites States of America [2], 2.11 in Canada [3] and 2.46 per 1000 live birth in United Kingdom (UK) [4].

Because the brain development continues during the first two years of life, CP can result from insults during prenatal, perinatal or postnatal periods such as: prematurity, low birth weight, intracranial hemorrhage, birth asphyxia and infection. [1], [5]. The diagnosis of CP is made based on the clinical pictures. The history of developmental delay, slow motor development, persistent infantile reflexes, hand preference earlier than 12 months of age all these observations are clues to the diagnosis of CP. A crucial point in the history is that the delay is not progressive. In this case, other neurological, metabolic, and genetic disorders must be considered [1].

Patients with CP commonly presented with spastic features, such as: increased muscle tone, increased deep tendon reflexes, scissoring of the lower limbs and toe walking. These features would represent 80% of patients with CP [6]. The other 15 % of patients would present with dyskinetic features such as abnormal limbs movements that increased with stress and disappear during sleep [7]. The least common presentation by 5% would be the ataxic type that affects the balance and coordination [7].

3. Methods

A six-year cohort data (2008-2015) was collected through chart-review of patients previously diagnosed with CP. The validity of CP diagnosis was questioned in the presence of atypical features in history, physical-exam or neuroimaging.

3.1 Tables

Table 1: Clinical Presentation

CP Type	*Percentage*
Spastic quadriplegic	12.1%
Spastic diplegia	3%
Dyskinetic	25.8%
Spastic paraplegia	25.8%
Hypotonia	19%

Table 2: The etiology

Clinical Presentation	*Confirmed Diagnosis*
Spastic quadriplegic CP	• Micro-Warburg Syndrome • Spinocerebellar Ataxia
Dyskinetic CP	• Myotonic Dystrophy • Cri du chat-variant • Mitochondrial Disorder • SOX deficiency • GLUT-1 deficiency

GLUT-1: Glucose Transporter Type 1 deficiency Syndrome

4. Results/Discussion

The total number of patients included in the study was fifty-nine patients: twenty-nine were females and thirty were males. Their age ranged between two and seventeen years. Their clinical presentations were summarized in table (1). The majority of patients were presented either as *Dyskinetic CP* (25.8%) or *Spastic paraplegic* CP (25.8%). Among those the etiology was identified in 13.5 % as in table (2). The most common metabolic abnormalities in cohort cases were abnormal amino acids by (27%), elevated lactate by (12%), abnormal liver function test (8%), elevated Hexosaminidase A-B ratio (7%), and elevated Arylsulfatase-B (5%).

Upon reviewing the neuroimaging of this cohort, different abnormal findings were reported. The most common findings

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were syringomyelia (12%), brain atrophy (10%), abnormal signal in basal ganglia (9%), cerebellar hypoplasia (7%), and white matter paucity (7%).

Patients who had abnormal laboratory results and neuroimaging underwent further genetic testing including Comparative Genomic Hybridization (CGH) and gene sequencing which confirmed the diagnosis. See table (3).

Table 3: The clinical and laboratory profile of diagnosed patients

Diagnosis	*Metabolic Workup*	*CGH/Gene sequencing*	*Neuroimaging*
Micro-Warburg syndrome	Normal	Normal/ Abnormal	Delayed myelination White matter paucity
Spinocerebellar ataxia	Elevated multiple amino acids	Normal/ Abnormal	Cerebellar hypoplasia
Myotonic Dystrophy	Elevated multiple amino acids	Abnormal	Abnormal signal in basal ganglia White matter paucity
Cri du chat variant	Elevated Hexosaminidase A	Abnormal	Syrinx White matter paucity
GLUT-1 deficiency	Decreased CSF glucose	Normal/ Abnormal	Cerebellar hypoplasia Syrinx
SOX deficiency	Normal	Abnormal	Cerebellar hypoplasia
Mitochondrial disorder	Elevated amino acids and lactate	Abnormal	Abnormal signal in basal ganglia

5. Conclusion

The presence of dyskinesia in clinical examination, in addition to abnormal findings in neuroimaging such as syrinx malformation, cerebellar hypoplasia, white matter paucity, and abnormal signal in basal ganglia may warrant further investigation for a disorder other than CP. Abnormal amino acids and other reported non-specific lysosomal enzyme profile might warrant further metabolic and genetic investigation. Normal CGH is not a stop sign to look further for into etiology

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