

# NEUROLOGIC MANIFESTATION OF ORGANIC ACADEMIA

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Inborn errors of organic acid metabolism are relatively recently recognized diseases with a wide spectrum of clinical signs and symptoms: ranging from asymptomatic, normal appearing children to death during first few days of life. In my presentation I will try to explain some of the most common clinical presentation of these disorder with stress on neurologic findings.

## **Organic acidemia usually have three clinical manifestations**

Severe neonatal form, Intermittent late-onset form and chronic progressive form. Recurrent coma, The main feature of these disorders is due to accumulation of toxic metabolites in Central Nervous system with direct effect on the function, while chronic accumulation of these materials may interfere with CNS development or cerebral metabolism leading to developmental delay.

### **Severe neonatal forms**

Following a symptom free interval of a few days from birth, poor sucking and difficult feeding appears in the newborn, followed by unexplained and progressive coma. Seizures may appear during the course of the disease and EEG may show a burst-suppression pattern. During this stage most infants have axial hypotonia with peripheral dystonia, choreoathetosis, episodic opisthotonus and some repetitive bicycling and boxing movements.

Associated biochemical abnormalities including metabolic acidosis, ketonuria and hyperammonemia also is usually present. The overall short-term prognosis with recent advances in medical care is improving. But later in life acute intercurrent episodes triggered by a stress often occur, which can be occasionally fatal. bulging fontanelle and cerebral edema may mimic CNS infection in these babies.

### **Intermittent late-onset forms**

Recurrent attacks of coma or lethargy with ataxia can occur in childhood or even in adolescence or adulthood. These episodes may be frequent, though in between these the child is entirely normal. These attacks are precipitated by conditions that enhance protein catabolism (trauma, infection etc).

Sometimes these episodes can lead to death or severe sequel. Seizure disorder is one of these sequels which is generalized in type with myoclonic seizure in infancy and childhood and later tonic-clonic and atypical absence seizures predominate.

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Also many of the survivors have acute or progressive extra pyramidal syndrome due to bilateral necrosis of basal ganglia.

### **Chronic progressive forms**

Non specific Developmental delay, hypotonia, muscular weakness, microcephaly and seizures are rarely the only revealing signs in organic acidemia without any acute presentation.

Seizures may become refractory to Anti Epileptic Drugs. In addition many asymptomatic or minimally symptomatic infants have been identified during tandem mass spectrometry newborn screening program. Cognitive deterioration associated with movement disorder such as dystonia or chorea may be caused by any form of organic aciduria.

**Keywords:** Organic Acidemia; Neurologic Manifestation; Clinical signs.