DI 23022.943 Dravet Syndrome

COMPASSIONATE ALLOWANCES INFORMATION	
DRAVET SYNDROME	
ALTERNATE NAMES	Severe Myoclonic Epilepsy Of/In Infancy; SMEI Syndrome; Epilepsy with Polymorphic Seizures; Polymorphic Epilepsy in Infancy; PMEI
DESCRIPTION	Dravet syndrome (DS) is a rare, genetic epileptic encephalopathy (dysfunction of the brain) with onset during the first year of life. Mutations of the SCN1A gene cause up to 80% of diagnosed cases of DS. Frequently referred to as a sodium channelopathy, this intractable epilepsy is characterized by unilateral (one-sided) clonic or tonic clonic (grand mal) seizures that are prolonged (> 5 minutes) or progress to status epilepticus (>30 minutes) and require emergency management. Myoclonic seizures, often called myoclonic jerks, are common. Over time seizures occur more frequently without obvious triggers, and resistant to treatment. Between one and four years of age, children develop other seizure types including atypical absence, eyelid myoclonia and non-convulsive seizures. All seizure types may be prolonged or lead to status epilepticus--a state of continuous seizure requiring emergency medical care. Children with DS typically experience poor development of language and motor skills, hyperactivity, and difficulty relating to others.
DIAGNOSTIC TESTING, PHYSICAL FINDINGS, AND ICD-9-CM/ICD-10-CM CODING	**Diagnostic testing:** Genetic testing for mutations within the SCN1A gene; EEGs. **Physical findings:** • Ataxia; • Dysarthria; • Intention tremor; and • Abnormal eye movement disorder. **ICD-9:** 345.1 **ICD-10:** G40

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PROGRESSION	Infants with Dravet syndrome appear normal at birth with most children showing signs and symptoms of this disorder during the first year of life. As children with Dravet syndrome get older, the degree of intellectual impairment appears to correlate with the frequency of seizures. The decline in cognitive function tends to stabilize after age 4. Children surviving into adolescence and adulthood are dependent on caregivers.
TREATMENT	Seizures in Dravet syndrome are difficult to manage, but can be reduced by anticonvulsant drugs. Some medications may aggravate seizures necessitating close monitoring of medication use by the claimant's medical source(s). Treatment with a ketogenic diet high in fats and low in carbohydrates has been of benefit in some cases.

SUGGESTED PROGRAMMATIC ASSESSMENT*

Suggested MER for Evaluation:

• Clinical history and examination that describes the diagnostic features of the impairment, and physical and cognitive findings;

• Imaging studies such as CT, MRI, or PET scans documenting structural changes in the brain;

• EEG reports measuring abnormalities in electrical activity in the brain; and

• Laboratory testing to rule out other causes (such as low or high blood sugar, low sodium, low magnesium or thyroid disorder) for seizures and for mutations in the SCN1A gene.

Suggested Listings for Evaluation:

DETERMINATION	LISTING	REMARKS
Meets	11.02	Listing level severity must be documented.
	111.02	Listing level severity must be documented.