CPSP
CANADIAN PAEDIATRIC SURVEILLANCE PROGRAM

Paediatric myasthenia

PROTOCOLS

Principal investigators

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Background

Paediatric myasthenia can be divided into two broad categories:

1) Myasthenia gravis (MG): This is an acquired **autoimmune** mediated disorder causing muscle weakness secondary to antibodies that bind to acetylcholine (ACh), MuSK (muscle specific kinase) or other receptors on postsynaptic muscle membranes, so that nerve impulses do not get through to the muscle.
2) Congenital myasthenic syndromes: Another form of myasthenia is congenital myasthenic syndrome, a rare **inherited** condition caused by structural defects of several types in the neuromuscular junction.

The main symptoms of myasthenia include rapid fatigue, hypotonia, muscle weakness (worst with repetitive activities), ptosis, diplopia, dysphagia, trouble feeding, respiratory difficulties, poor head control in infants and delayed development.

Paediatric myasthenia is much rarer than the adult version of this condition and patients are frequently symptomatic for years before the specific diagnosis is made. As there are effective treatments and often curative measures for the autoimmune form, such as cholinesterase inhibitors, corticosteroids, plasmapheresis, intravenous immunoglobulin or thymectomy, early detection and diagnosis is in the child's best interest. In addition to the substantial physical and emotional toll of having any chronic disease, myasthenic patients have to endure the general lack of awareness of the condition.

As few dedicated published series are available, national collaboration amongst medical professionals is critical to obtain a realistic assessment of the incidence and impact of paediatric myasthenia. It is anticipated that the CPSP study will lead to documentation of the burden of illness and inform on best practices.

Methods

Through the established methodology of the CPSP, over 2,500 paediatricians and paediatric subspecialists will be actively surveyed on a monthly basis for identified cases of paediatric myasthenia.

Objectives

1) To increase awareness of paediatric myasthenia amongst paediatricians.
2) To reinforce knowledge of myasthenia diagnosis and management.
3) To ascertain the incidence of paediatric myasthenia.
4) To determine current treatments offered to myasthenic children across Canada, while observing geographical trends.

Case definition

Report any child less than 18 years of age with at least one of the following clinical features:

- fluctuating ptosis (unilateral or bilateral) and/or
- fluctuating extraocular muscle weakness (unilateral or bilateral) and/or
- history of skeletal muscle weakness or fatigue

AND any of the following supportive tests:

- Tensilon™ test (edrophonium) (or other acetylcholinesterase inhibitor) demonstrating reversal of weakness
- elevated acetylcholine receptor or MuSK (muscle specific kinase) antibody levels
- abnormal nerve conduction studies (demonstrating defect in neuromuscular junction transmission) or single fiber EMG

Exclusion criteria

- Underlying primary muscle disease
- Underlying metabolic disease
- Transient neonatal myasthenia

Duration

January 2010 to December 2011

Expected number of cases

The estimated incidence for this study is approximately 60 new cases per year.

Ethical approval

University of Alberta Health Research Ethics Board

Analysis and publication

An annual interim analysis of the collected data will be done. Dissemination of completed study results will be submitted for publication to appropriate peer-reviewed journals and presented at national and international scientific meetings.

Bibliography

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