Introduction

X-linked Myotubular Myopathy presents in newborns as profound weakness and generalized hypotonia. This X-linked inheritance of this disease is associated with the most severe clinical presentation. Prognosis depends on the extent of the disease and varies with each individual. The mode of inheritance plays a factor as there are multiple autosomal dominant and autosomal recessive forms.

The X-linked form is a non-progressive neuromuscular disorder, but despite this, this condition is ultimately fatal with infants having a life expectancy ranging from weeks to months.

Unfortunately there is not a cure for this disease and treatment remains prognostically supportive, involving a multi-disciplinary health care team.

The family should be counseled and informed about the natural course of the disease and realistic expectations should be sought. Furthermore, counseling should be given about future pregnancies and risks of recurrence.

Abstract

We will be presenting a case on the neonatal, diagnosis, and management of a 2230 gram male infant born via C-section at 35 weeks gestational age.

Our patient presented to the NICU in respiratory distress/failure management of a newborn. The patient was intubated and transferred to the NICU for further NICU management. His clinical presentation, autonomic function tests, head CT, MRI, respiratory function tests, and echocardiography were within normal limits. His smoking history, parental consanguinity, and family history were unremarkable.

After the diagnosis was made, the news was delivered to the parents and the treatment options and the natural progression of the disease were explained to them. It was then the decision was made to sign a DNR and withdraw ventilatory support and allow our patient to breathe on his own. Our patient died of cardiorespiratory failure three weeks afterwards.

Results

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Conclusions

X-linked Myotubular Myopathy has an incidence from 2-5/100,000. It is an inherited disorder that can be X-linked (most severe clinical presentation), autosomal recessive, or autosomal dominant.

Clinical presentation and severity of X-linked Myotubular Myopathy is variable but can have these characteristics:

- Profound weakness/hypotonia
- External ophthalmoplegia
- Respiratory failure
- Thinning of ribs
- Undescended testes

Our diagnosis was confirmed by muscle biopsy and genetic testing identifying a mutation in the myotubulin gene on chromosomes Xq28. Biopsy usual shows centrally placed nuclei with glycogen accumulation. This case was unique as it presented with an unaffected mother.

Treatment, unfortunately remains predominantly supportive, usually requiring ventilation support.

References


