

NEONATAL DIAGNOSIS AND TREATMENT OF STIFF BABY SYNDROME: A CASE REPORT

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BACKGROUND AND IMPORTANCE



STIFF syndrome is a rare disease with genetic mutation **Cr 5 GLRA1**. It is characterized by a neurological disorder with stiffness and muscle spasms, which affects the quality of life.

AIM AND OBJECTIVES

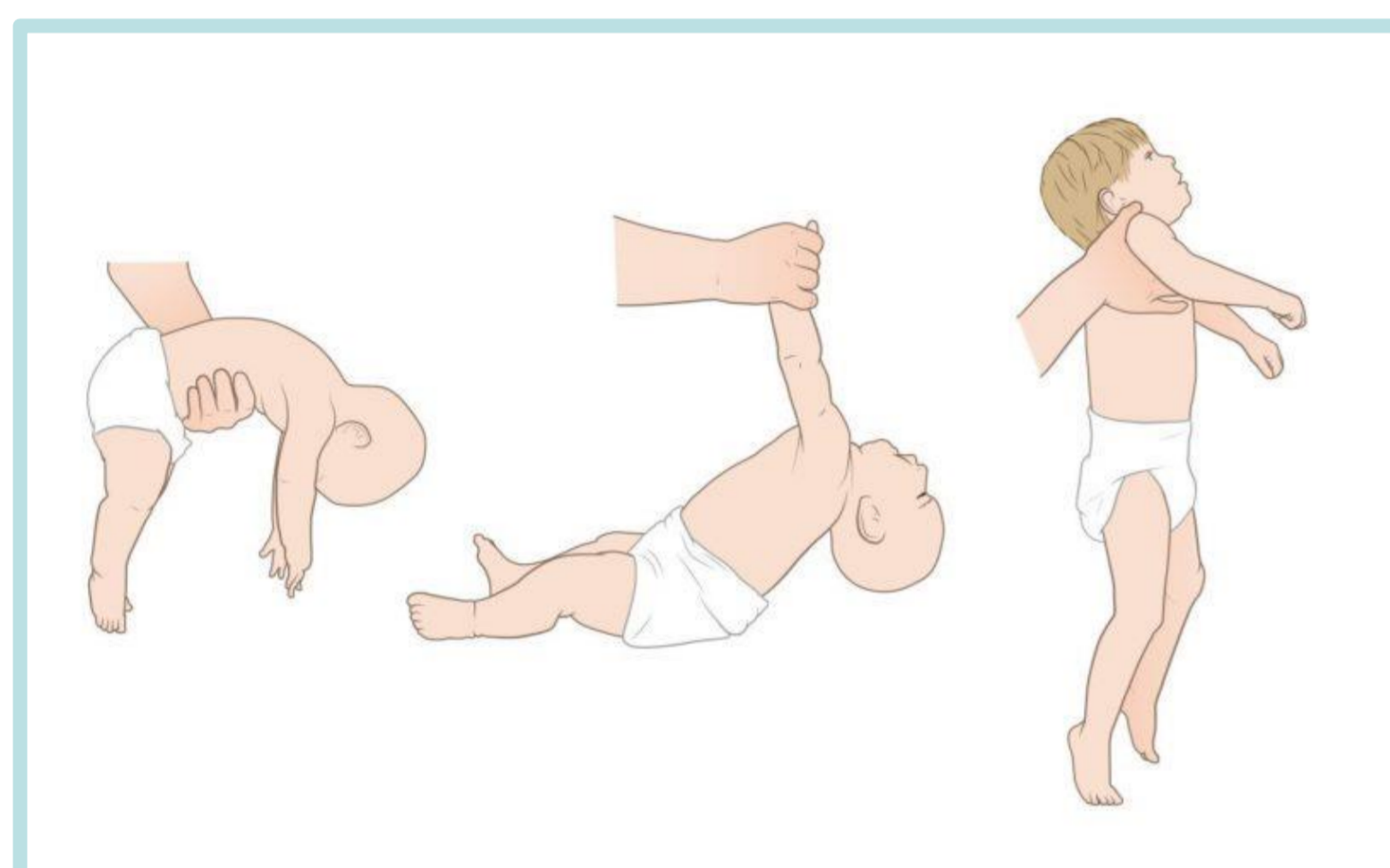
Describe the diagnostic and therapeutic management of a neonatal patient with Stiff syndrome.

MATERIALS AND METHODS

Literature review of cases described with similar clinical features by the **pharmacy and neonatology** service of a tertiary hospital. Tests were requested for differential and confirmatory diagnosis (whole genome sequencing). The Pharmacy Service collaborated in the search for a possible effective treatment and in adapting it to a paediatric patient.



RESULTS



Premature patient (41+2) hospitalized the 16 of August 2022 in a tertiary hospital due to respiratory distress and abnormal neurological signs, followed by a hypertonic seizure with generalised rigidity.

A bolus of midazolam 0.1 mg/kg was administered without improvement, followed by phenobarbital 3 mg/kg/24h without clinical response.

After negative tests, the genetic study detected an alteration of the **GLRA1 gene** in the patient, and a heterozygous mutation in mother. A metabolic study was performed, detecting elevated levels of **glutamic acid** (1)

A therapeutic trial was started the 28 of August with oral **Clonazepam at 0.1 mg/kg every 8 hours** (1). As this was a compounding preparation, the pharmacy prepared the suspension at a concentration of 0.1 mg/ml from 2 mg tablets (2).



Due to the improvement in stiffness and hypereplexia since the start of treatment, clonazepam was maintained at discharge, and continues being active at 0.3 mg/kg/8 hours. At follow-up at 11 months of age, the patient is in good general condition. **The condition has attenuated, with less startle and reflexes.**

CONCLUSION AND RELEVANCE

Stiff syndrome is a disease that is **difficult to diagnose and to treat** due to its **low prevalence**. The favourable clinical response after starting treatment with clonazepam should be highlighted. The preparation of a pharmaceutical formulation from the Pharmacy Service allowed to individualise the dose according to the patient's weight and clinical evolution.

BIBLIOGRAPHY

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