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Commentary

Therapeutic Implications in the Treatment of Phenotypic Mutations

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ABOUT THE STUDY

Mutations in the Neuraminidase (*NEU1*) gene cause Sialidosis, also known as Mucolipidosis Type 1, a rare autosomal recessive Lysosomal storage condition. Over 40 harmful *NEU1* mutations have been identified. The age of disease onset and the severity of symptoms are impacted by the many mutations that cause varying levels of residual Neuraminidase function. Sialidosis type 1 presents with a milder Phenotype and onset at a later age; Sialidosis Type 2. One in every 4.2 million live births results in Sialidosis. Sialidosis type 1 often manifests itself in the second or third decade of life. Since these two symptoms, together with Ataxia, Hyperreflexia, and visual loss, are typically present in type 1, the term "Cherry Red Spot Myoclonus" is frequently used to describe it. There may also be progressive Myoclonic Epilepsy. Light sensitivity is typically more a sign of other types of progressive Myoclonic epilepsy, like Gaucher's disease, Lafora Disease, or Ceroid-Lipofuscinosis. Patients with Sialidosis Type 1 can have resting or action Myoclonus, which can be sensitive to Tactile and Aural Stimuli. The Myoclonus can manifest axially or in an appendicular pattern [1].

Pathogenic mutations in the *NEU1* gene and decreased Neuraminidase activity in cultured fibroblasts and Leucocytes are required for the diagnosis of Sialidosis. Patients with Sialidosis Type 1 might Pheno typically differ greatly from one another due to the wide variation in illness symptoms and signs, particularly of the Myoclonus. Sialidosis patients have not yet been the subject of thorough eye movement evaluations. A mild downbeat Nystagmus on lateral gaze and a subtle gaze-evoked Nystagmus with a few upbeat beats on far upgaze were both detected during an ocular motor evaluation [2].

Both Saccadic smooth pursuit and Saccadic Hypermetria were seen. Due to his poor visual acuity, attempts to record eye movements failed to yield any useful information [3].

A Saccadic horizontal smooth pursuit gain greater than vertical smooth pursuit gain, intermittent horizontal and vertical Saccadic Hypometria, and a prolonged vertical Saccade Latency were all found during an ocular motor examination. Optokinetic Nystagmus was normal in the horizontal plane but only in the slow phases in the vertical plane indicating the potential onset of Saccadic Gaze Palsy. Convergence insufficiency and a slight right sixth nerve paresis were also present. Recordings of eye movements demonstrated Saccadic Hypometria, Saccadic Horizontal and vertical slowing (confirming a Saccadic Gaze Palsy, most likely caused by involvement of brainstem burst neurons), and saccadic horizontal and vertical smooth pursuit [4].

Recessive Sialidosis Type 1 is a rare disease. Pathogenic variants of the *NEU1* gene that cause neuraminidase enzyme deficiency impair the metabolism of Sialic acid on Oligosaccharides and Glycoproteins. Sialic acid-Rich Macromolecules build up as Lipofuscin-Rich inclusions in the Cytoplasm of cells in various target tissues, leading to cellular dysfunction and disease manifestations with varying degrees of severity depending on the enzyme's remaining activity [5].

Myoclonus, ataxia, seizures, and a variety of visual complaints make up the classic presentation of Sialidosis type 1.

CONCLUSION

Not every patient exhibits the full range of the disease, even though cases with an earlier age of onset more frequently have a classical presentation. As a result, it is more challenging to diagnose this already uncommon condition, especially when clinicians are unfamiliar with how it manifests. Patients and their families are burdened by a delayed diagnosis. By presenting these three Sialidosis Type 1 patients, who are all well-characterized, we hope to raise clinician awareness of this condition by thoroughly documenting phenotypic variability and treatment responses. Myoclonus was significantly triggered by movement in all three cases, though it was also noticeable at rest. Given its therapeutic implications, Myoclonus in these patients was responsive to alcohol, as was to be expected. These patients may benefit from conventional methods using Benzodiazepines and Antiepileptic Medications, which are typically combined. However, Sodium Oxybate can also be considered when symptoms are resistant to first-line treatments. In the past, Cataplexy in Narcolepsy has been treated with SBX, a Sodium Salt of-Hydroxybutyrate. While

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treating Alcohol-Responsive movement disorders like tremor, Post-Hypoxic Myoclonus, Myoclonus Dystonia, and Spasmodic Dysphonia, this medication has been shown to be effective. In every one of our patients although one had to stop using it due to side effects, the treatment significantly increased one's quality of life.

REFERENCES

1. Wolach O, Stone RM. Optimal therapeutic strategies for mixed phenotype acute leukemia. Curr Opin Hematol. 2020;27:95–102.
2. Weinberg OK, Arber DA. Mixed-phenotype acute leukemia: Historical overview and a new definition. Leukemia. 2010;24:1844–1851.
3. Matutes E, Pickl WF, Van't Veer M, Morilla R, Swansbury J, Strobl H, et al. Mixed-phenotype acute leukemia: Clinical and laboratory features and outcome in 100 patients defined according to the WHO 2008 classification. Blood. 2011;117:3163–3171.
4. Yu J, Li Y, Xing H, Pan Y, Sun H, Wan D, et al. Clinical Characteristics and Outcome of Biphenotypic Acute Leukemia: 10 Case Reports and Literature Review. Cancer Manag Res (2019) 11:9297–306.
5. Kajal B, Chang H. Acute myeloid leukemia with myelodysplasia-related changes demonstrating mixed-lineage phenotype. Blood 2016; 128: 1663. 2017/01/18.