What is HIST1H1E (HNDS)

HIST1H1E gene encodes one of the histone linker proteins in human. The basic building block for a structure called chromatin. The chromatin is essential part of common structure called Chromosome. The name of HIST1H1E has been described differently over the time such as H1.4 and H1E. The formal name is now called H1-4 in scientific literature.

What causes variations in the HIST1H1E Gene

The variations or mutations (changes of DNA) found in patients are new to patients, we typically referred this as “de novo” event in the genetics resorts and the same mutations were not found in either parent. The cause or reason why and how these mutations occurred is not well understood in most of cases. In most or all cases, they just happened randomly.

What are the Symptoms

The full spectrum of clinical symptoms and problems in patients with HIST1H1E mutations is still not fully delineated. Based on the information we learned from a limited number of patients with HISTH1E(H1-4) mutations, the following may be seen:

- Low muscle tone or hypotonia at birth;
- Delayed developmental milestones and mental delay;
- Language developmental delay;
- Eye and vision problems;
- Autism spectrum disorder;
- Unusual or dysmorphic;
- Congenital heart defect;
- Bone related issues;
- Anxiety;
- ADHD;
- Sensory issues;
- Recurrent infections;
- Others but probably rare such as seizure, big head, overweight, GI issue, etc.

Who can receive a HIST1H1E diagnosis

Any individuals with significant intellectual disability, developmental delay, and autism spectrum disorders should be evaluated for HISTH1E/H1-4 mutations by genetic test and could receive the HIST1H1E diagnosis if the mutation is positive.

How is HIST1H1E diagnosed

Genetic or DNA test is most reliable way to make the diagnosis.

Is there a cure?

No known treatment that can cure HIST1H1E now.

What are the treatments

Current treatments are symptomatic care and recreational/educational interventions.

What is the prognosis

The exact prognosis at different ages is still not known. Overall, HIST1H1E patients are doing well from the patients we learned and cared so far. Some of them are living at @ ~50 years of age. They are medically well and healthy but do have mental delay and behavioral issues etc. The signs of premature aging are seen in some patients, but it is not clear how common is this.

What kind of equipment might my child with HNDS need

No specific equipment is needed. But some physicians may recommend learning device and others to help some symptoms.

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