Common Questions

DYRK1A SYNDROME

WHAT IS DYRK1A?

DYRK1A is another name for the dual specificity tyrosine phosphorylation regulated kinase 1A gene. It is located on chromosome 21 in the q22.13 region and provides instructions for making a kinase enzyme. The DYRK1A enzyme is important in the development of the nervous system.

WHAT IS DYRK1A SYNDROME?

DYRK1A Syndrome is described by a specific set of signs and symptoms. These signs and symptoms can include; intellectual disability, speech impairment, feeding difficulties at birth or during infancy, epilepsy or history of seizures, autistic traits, reduced head size (microcephaly), and certain facial characteristics.

WHAT CAUSES DYRK1A SYNDROME?

DYRK1A Syndrome is caused by genetic changes that occur in the DYRK1A gene which affects the function of the DYRK1A protein. Genetic changes most often happen very early in a pregnancy, during or soon after conception. These genetic changes are often called variants or mutations. This gene has an important role in regulating the function of many other proteins in the cell. However, some variants in the DYRK1A gene may not affect the protein.

HOW IS DYRK1A SYNDROME DIAGNOSED?

DYRK1A changes can be found with molecular genetic testing. Chromosomal microarray analysis (CMA) can detect large deletions or duplications that affect the DYRK1A gene. If a change is too small to be detected there are a few other methods. Multigene panels are used to look for changes in groups of genes related to a specific symptom. The DYRK1A gene is included in multigene panels for intellectual disability (ID), microcephaly, autism & epilepsy. Whole exome or genome sequencing can also detect changes in the DYRK1A gene. A geneticist can explain the results of these tests.

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WHAT VARIANTS ARE FOUND IN DYRK1A SYNDROME?

The variants reported in current research articles cause a loss of functional DYRK1A protein from one of the two copies of the gene. There are many different variants that can lead to a loss of function to the protein. These could be variants that lead to an absence of DYRK1A expression, or to a protein missing one part of its sequence, and sometimes to a protein with a very subtle change in its sequence, but this small change is sufficient to impair its activity.

DO DIFFERENT VARIANTS CAUSE DIFFERENT SYMPTOMS & SEVERITY?

There is no evidence to date that different variants also present different symptoms or severity. It is possible though not certain that variants that only partially affect the DYRK1A activity if they exist, may lead to less severe clinical manifestations. In contrast to other syndromes that result in forms of intellectual disability, there are similarities among individuals carrying a pathogenic variant in DYRK1A. We hypothesize that other factors, from genetic or environmental origins, may influence the clinical manifestations and the severity of the syndrome.

IS 21Q22.13 DELETION THE SAME?

Yes, DYRK1A is the gene responsible for the symptoms observed in the individual carrying a deletion of this 21q22.13 region. A deletion may also be described as a partial deletion or micro deletion. Individuals with 21q22.13 deletion and anomalies/variants in DYRK1A present the same clinical manifestations.

HOW IS DYRK1A SYNDROME CONNECTED TO DOWN SYNDROME?

The DYRK1A gene is located on chromosome 21. Typically, people have two copies of chromosome 21 but people with Down Syndrome have three copies of chromosome 21. Having three copies of the chromosome also causes them to have three copies of the DYRK1A gene and then makes too much of the DYRK1A protein. You might see research that is trying to inhibit or decrease the amount of DYRK1A protein. Decreasing the DYRK1A protein wouldn't be helpful for people with DYRK1A Syndrome because the mutations found in DYRK1A Syndrome cause one of the gene to not function properly and then creates too little of the DYRK1A protein.

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WHAT SHOULD I EXPECT FOR MY CHILD'S DEVELOPMENT?

Each person with this diagnosis is unique and may not be susceptible to developing all the features that have been reported. It is common to present with a delay in psychomotor development, significant difficulties with communication and language, and epileptic seizures in early childhood, which may or may not continue into adolescence and adulthood. Some behavioral issues such as anxiety or stereotypic behavior, often referred to as stimming and a common trait among autistic individuals may occur. They may have feeding challenges beginning at birth with difficulty coordinating a suck, swallow, and breath pattern that can progress with strong preferences to different textures.

IS THERE A CURE?

There is currently no cure for DYRK1A Syndrome.

WHAT ARE THE TREATMENTS?

Early intervention and regular therapies such as speech, occupational, and physical therapies can help manage symptoms and also help reach developmental milestones. Medical conditions related to DYRK1A Syndrome are treated by multiple different specialists.

WHAT IS THE PREVALENCE OF DYRK1A?

It is difficult to know the exact number of cases because a formal registry does not exist. However, according to large-scale studies, around 0.3% to 0.5% of individuals with intellectual disability are likely to carry a genetic anomaly (variant) in DYRK1A based on genetic testing. The number of individuals identified will increase in the coming years as genetic testing becomes more utilized. As an example, there are more than thirty affected individuals identified in France and we expect ten more identified per year.