DYRK1A Gene

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Dual Specificity Tyrosine Phosphorylation Regulated Kinase 1A

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1. Normal Function

The *DYRK1A* gene provides instructions for making an enzyme that is important in the development of the nervous system. The DYRK1A enzyme is a kinase, which means that it adds a cluster of oxygen and phosphorus atoms (a phosphate group) to other proteins through a process called phosphorylation. Phosphorylation of proteins helps to control (regulate) their activity.

The proteins whose activity the DYRK1A enzyme helps regulate are involved in various processes in cells, including cell growth and division (proliferation) and the process by which cells mature to carry out specific functions (differentiation). In nerve cells (neurons), the DYRK1A enzyme is involved in the formation and maturation of dendritic spines from dendrites. Dendrites are specialized extensions from neurons that are essential for the transmission of nerve impulses. Dendritic spines are small outgrowths from dendrites that further help transmit nerve impulses and increase communication between neurons.

2. Health Conditions Related to Genetic Changes

2.1 Autism Spectrum Disorder

At least 11 *DYRK1A* gene mutations have been identified in people with autism spectrum disorder (ASD), a varied condition characterized by impaired social skills, communication problems, and repetitive behaviors. Most people with ASD associated with *DYRK1A* gene mutations also have other signs and symptoms. In some cases, they have a particular combination of additional features, including intellectual disability, speech problems, anxiety, and an unusually small head (microcephaly). Other signs and symptoms that may occur in these individuals include recurrent seizures (epilepsy), characteristic facial features, weak muscle tone (hypotonia), foot abnormalities, and walking problems (gait disturbance). This pattern of signs and symptoms is sometimes called *DYRK1A*-related intellectual disability syndrome.

DYRK1A gene mutations result in loss of the DYRK1A enzyme or an enzyme that does not function properly. Impaired or absent DYRK1A enzyme function likely leads to abnormal regulation of gene expression and disrupts proper neural development. However, the specific relationship between *DYRK1A* gene mutations and the signs and symptoms of ASD, as well as the other features that may occur in people with these mutations, is unclear.

3. Other Names for This Gene

- dual specificity tyrosine-(Y)-phosphorylation regulated kinase 1A
- dual specificity YAK1-related kinase
- DYRK
- DYRK1
- HP86
- MNB
- mnb protein kinase homolog hp86

- MNB/DYRK protein kinase
- MNBH
- MRD7
- protein kinase minibrain homolog
- serine/threonine kinase MNB
- serine/threonine-specific protein kinase

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