# PURA And 5q31

*https://pubmed.ncbi.nlm.nih.gov/28448108/*

Clinical characteristics:

PURA

-related neurodevelopmental disorders include

PURA

syndrome, caused by a heterozygous pathogenic sequence variant in

PURA

, and 5q31.3 deletion syndrome, caused by a genomic 5q31.3 deletion encompassing all or part of

PURA.

PURA

-related neurodevelopmental disorders are characterized by moderate-to-severe neurodevelopmental delay with absence of speech in most and lack of independent ambulation in many. Early-onset issues can include hypotonia, hypothermia, hypersomnolence, feeding difficulties, excessive hiccups, recurrent central and obstructive apneas, epileptic seizures, abnormal nonepileptic movements (dystonia, dyskinesia, and dysconjugate eye movements), and abnormal vision. Congenital heart defects, urogenital malformations, skeletal abnormalities, and endocrine disorders occur, but are less common.

Diagnosis/testing:

The diagnosis of a

PURA

-related neurodevelopmental disorder is established in a proband with either a heterozygous

PURA

pathogenic sequence variant (90% of affected individuals) or a nonrecurrent deletion of 5q31.3 that encompasses all or part of

PURA

(10%).

Management:

Treatment of manifestations:

Ongoing routine care by a multidisciplinary team. Treatment and/or therapy for developmental delays; neurologic findings (hypotonia, seizures, abnormal movements); feeding difficulties; apnea;

visual impairment; and malformations of the heart, urogenital tract, and skeleton.

Surveillance

: Long-term follow up to assess psychomotor development, seizures or suspected seizures, vision, feeding for dysphagia, and musculoskeletal complications (hip dysplasia and scoliosis).

Genetic counseling:

PURA

-related neurodevelopmental disorders, caused by either a heterozygous

PURA

pathogenic sequence variant or a 5q31.3 deletion encompassing all or part of

PURA

, are inherited in an autosomal dominant manner. In almost all probands with a

PURA

pathogenic sequence variant the sequence variant is

de novo

; to date, all reported 5q31.3 deletions have been

de novo

. For parents of an affected child, the risk to future pregnancies is presumed to be low, as a

de novo

genetic alteration involving

PURA

is most likely in the proband. However, parents of an affected child may wish to consider prenatal testing or preimplantation genetic testing, as risk may be greater than in the general population owing to the possibility of parental germline mosaicism (estimated empirically at <1%).