

PITT-HOPKINS SYNDROME

CAUSES OF PITT-HOPKINS SYNDROME

Pitt-Hopkins Syndrome is caused by a deletion of or a mutation in the TCF4 gene on chromosome 18q21.2.

It controls the activity of many other genes and plays an important part in how the brain develops and functions.

Free Pitt Hopkins app available on Windows, Android and IOS.

www.pitthopkins.org.uk

SEVERE
INTELLECTUAL
DISABILITY
DELAYED MOTOR
DEVELOPMENT

NON VERBAL OR
LIMITED SPEECH
POSSIBLE SEIZURES

DEEP-SET EYES
DISTINCTIVE EYE
BROW SHAPE

BREATHING
ANOMALIES
CONSTIPATION

SHORTER
IN HEIGHT
FOETAL PADS ON
FINGERS AND TOES

WIDE MOUTH
CUPID BOW TOP LIP
FULL BOTTOM LIP

IMPACT OF PITT-HOPKINS SYNDROME

People with Pitt-Hopkins Syndrome will never lead independent lives and most will need full-time care even at night as many have sleep problems.

Some will never walk and most do not talk.
Some can communicate with AAC devices or limited signing but many do not have access to this.

Most will need help with all aspects of personal hygiene but many will learn to feed themselves.
Some will achieve a degree of continence.

Some will have ongoing problems and pain with constipation often needing a hospital admission.

The constipation is usually treated with medication and diet and the careful monitoring of bowel movements. Some have problems with reflux.

Breathing abnormalities with intermittent periods of over-breathing and breath holding are not present in every person and may not appear until mid-childhood. The breathing anomalies are very mild and almost insignificant in some but in others the hyperventilation and apnoea can severely affect their lives. Acetazolamide helps some with the breathing anomalies but has no effect on others.

HOPES AND ASPIRATIONS

An understanding of what causes the breathing anomalies and how to treat them is very important.

A better knowledge of the chronic constipation and pain that many with Pitt-Hopkins Syndrome suffer.

A greater understanding of the autonomic system in people with Pitt-Hopkins Syndrome and its effect on their daily lives, not only to help with the severe constipation and breathing anomalies, but on other aspects of a dysfunctional autonomic system.



Pitt Hopkins UK

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