Pallister-Hall Syndrome (PHS)

By George William Helon

Introduction

These days it is commonplace for genetic disorders and syndromes to be diagnosed within the first few weeks of an infant's life. However some 20 to 30 years ago reaching a diagnosis was a difficult task. Patients could present with, or suffer from the effects of a single, or a multitude of complex and varied symptoms, abnormalities and malformations that were in some cases quite evident, or in others cloaked for a number of days, weeks, months, or, as in my case – years. In June 2001 I was diagnosed with Pallister-Hall Syndrome (PHS); I was 35 years of age.

What is PHS?

Pallister-Hall Syndrome (PHS) is an extremely rare genetic disorder that may be apparent at birth; it is a multiple congenital anomaly syndrome, not a disease. PHS follows a pattern of genetic inheritance in families called autosomal dominant inheritance. The disorder can be both sporadic or inherited. At this time (February 2002) there are approximately only 100 known diagnosed cases of PHS worldwide. I am only the second individual diagnosed with PHS in Australia.

What causes PHS?

Pallister-Hall Syndrome (PHS) is a genetically transferable (inheritable) disorder that is caused by an alteration (or mutation) to the gene GL13 (glee three) which is located on the short arm of the seventh (7^{th}) chromosome in the region known as 7p13.

Symptoms and Features

Those symptoms, findings, abnormalities, malformations and features associated with, and peculiar to Pallister-Hall Syndrome (PHS) can vary greatly in both range and severity from patient to patient and can affect individuals to different and varying degrees. The central and most significant feature of any diagnosis of PHS is the presence of a Hypothalamic Hamartoblastoma (HH) which may, or may not cause any health problems. A HH is a benign, non-cancerous malformation, or brain tumor of the, or in the Hypothalamic region of the brain that can cause deficiencies in one or more hormones, seizures and precocious puberty. Some of the other prominent features and conditions peculiar to PHS include: the presence of a "Y"-shaped metacarpal or metatarsal bone and extra fingers and/or toes; syndactyly; polydactyly (both central and postaxial); bifid or cleft epiglottis; imperforate anus; micropenis; pituitary dysfunction and craniofacial malformations. Secondary features are both numerous, complex and varied.

Who Can Get PHS?

Although statistically males seem to be more affected than females, Pallister-Hall Syndrome (PHS) is not specific to either gender, nor panethic, because the gene responsible for the disorder is found on chromosome 7. PHS is found in persons of all nations.

Treatment for PHS

Generally, there is no specific treatment for Pallister-Hall Syndrome (PHS) in its entirety because it is an extremely rare multiple congenital anomaly syndrome. However, some of

those characteristic symptoms, conditions, abnormalities or malformations peculiar to PHS can be treated individually by hormone supplements and/or replacements, by surgery, or by other means. In all cases though, those diagnosed with PHS should have periodical examinations in order to monitor the state, that is the size, condition and/or composition of the Hypothalamic Hamartoblastoma. Periodical monitoring should be by way of Magnetic Resonance Imaging (MRI).

Children and PHS

Those couples contemplating having children obviously would be concerned about passing the disorder onto their off-spring. Where one parent is affected by Pallister-Hall Syndrome (PHS) there is a fifty-per-cent (50%), or 50/50 chance that the child could inherit the chromosome that does not contain an altered GL13 gene. There is no evidence to suggest that the severity of inheritance is lessened when a parent is "mildly" affected.

Further Information

To find out more about Pallister-Hall Syndrome (PHS): its causes; symptoms; abnormalities; malformations; findings; diagnosis; treatment and for a full explanation of difficult words, terms and abbreviations specific to PHS, **go to my Facebook page at: https://www.facebook.com/Pallister.Hall.Syndrome**
or contact me on ghelon@yahoo.com.au.

Footnote

Between 3-7 December 2001 I attended the National Institutes of Health (NIH) National Human Genome Research Institute (NHGRI), Bethesda, Maryland in the United States of America. The NIH is one of the world's foremost facilities for biomedical research. After detailed work-ups, evaluations and testing my diagnosis was confirmed by Dr Leslie G. Biesecker, M.D. who is deemed the world's leading authority on PHS. Dr Biesecker is a Certified Clinical Geneticist and Pediatrician.