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SPECIAL POINTS OF INTEREST:

- New evidence that IC/BPS/CPPS is inherited!
- Visit our new Website!
- WWW.CHB-genomics.org

Genetics Study of IC/PBS/CPPS Newsletter

Further Evidence that PBS/IC is a Dominantly Inherited Disorder



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Painful bladder syndrome/interstitial cystitis (PBS/IC) is a debilitating syndrome characterized by chronic pelvic pain (severe to excruciating), urinary frequency (can be over 50 urinations per day), and urinary urgency (can cause sleep deprivation).

To date, the cause of

PBS/IC remains unknown. The idea that PBS/IC is a bladder disease is in dispute, there are arguments that PBS/IC affects the entire the body. The increased incidence of other painful conditions in patients with PBS/IC and their family members (irritable bowel syndrome, migraine headaches, fibromyalgia, and vulvodynia) supports this idea that PBS/IC is a systemic (throughout the body) disease.

Understanding the true nature of PBS/IC will be essential to developing effective therapies. Several research studies have supported the idea of an inherited risk for PBS/IC. It has been shown that adult female relatives of PBS/IC patients have an increased risk to have PBS/IC symptoms over the general population. Twin studies showed

that 67% of identical twins had PBS/IC compared to a 0% in fraternal twins. Because identical twins share more of their genes than fraternal twins this points to a significant genetic susceptibility for PBS/IC. Previously, we reported the first example of familial clustering of PBS/IC. Subsequent investigation led to the identification of 35 families with more than one case of PBS/IC in Bulgaria. Six of these families were studied for identifying the underlying genetic defect. Our preliminary data confirms that at least five different genes may result in PBS/IC with an autosomal dominant inheritance pattern in the Bulgarian population.

Historically, PBS/IC cases in the US and Canada were thought to be sporadic or have few affected family members. With building evidence for a familial risk for PBS/IC to first degree relatives of an affected person in the Bulgarian population, a group of patients from North America (US and Canada) was recruited. This population also showed an increased number of families with multiple affected members; approximately 65% of families have more than one PBS/IC-affected member and several families have three generations of affected members. Affected individuals include both males and females and represent many ethnicities. These findings advance and strengthen the hypothesis that, contrary to conventional belief, PBS/IC is an inherited disorder.

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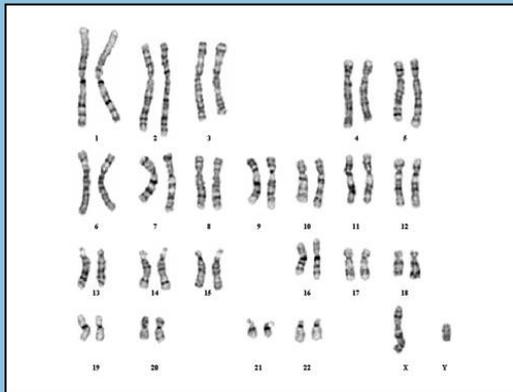
Visit us on the web!

www.chb-genomics.com

Then click on Interstitial Cystitis



We'd love to hear from you!



Karyotype = picture of 23 pairs of chromosomes found in a man. Chromosomes pairs are numbered by size from largest to smallest

NOTE: X & Y sex chromosome indicating a male

Why do you look like your father? Is your hair curly like your mother? These features are inherited or passed down from our parents. Our genes determine all of our features and body functions. These genes are considered our genetic blueprint or instructions. Our genes are grouped in packets called **chromosomes**.

What is Dominant Inheritance?

Each person has **46** chromosomes that come in pairs (23 pairs). One chromosome from each pair is inherited from your mother and the other from your father. This is how our parents and grandparents have passed their genes down to the next generation. How our genes are expressed in our family is called the **inheritance pattern**.

We know that chromosome pairs numbered are 1-22 by size (1 largest-22 smallest). These chromosomes are called **autosomes**. The 23rd pair of chromosomes determine our gender. **Males** have an **X & Y chromosome** and **females** have **X & X**. This last pair is called the **sex chromosomes**.

From our research, we believe that PBS/IC follows an Autosomal Dominant inheritance pattern in most families. A disorder may be dominantly inherited if at least one person in each generation has disease symptoms. As each person has 2 copies of his/her genes, dominant disorders only require **1 of the 2 copies to be altered or changed** causing disease symptoms. This is opposed to recessive inheritance in which both copies of the same gene are altered to cause disease.

Autosome: gene is on chromosome 1-22 not on sex chromosomes, X or Y

Dominant: 1 copy of a gene is changed (mutated) & causes disease symptoms