

East Hosts West in a Grand Way

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*The face of our future not only comes from research in the USA,
but from research around the world.*

Although they had never hosted a national PWS conference let alone an international one, Taiwan, our host county, did a phenomenal job creating a spectacular 7th International Prader-Willi Syndrome Conference. Having the conference in an Asian country brought in many presenters and attendees that have not had the opportunity to participate before. We also had a strong share of USA involvement as organizers and presenters. The Caregiver's program was exceptionally well attended, and Dr. Li Ping Tsai told me afterwards, that it has given their families in Taiwan the knowledge and motivation to begin the process of establishing supportive living homes in their country.

The following are glimpses of a few of the 29 oral reports and 36 poster presentations from this conference. The entire report must be heard and/or read to understand the conclusions. This is non-peer reviewed research (a process required by medical journals). The full abstracts, including more complete credits are currently posted on the PWSA (USA) web site at www.pwsausa.org and on the IPWSO Taiwan website at www.ipwso.org.

INTERNATIONAL PRADER-WILLI SYNDROME ORGANISATION 7TH SCIENTIFIC CONFERENCE MAY 20-21, 2010, TAIPEI, TAIWAN

Co-chairs: Suzanne B Cassidy, M.D. & Leopold M.G. Curfs, PhD

HYPOTONIA IN NEONATES WITH PWS IS NOT ALWAYS SEEN AT BIRTH BUT BECOMES EVIDENT AFTER THE FIRST DAYS -- Susanne Blichfeldt -- Denmark

Hypotonia and other typical symptoms in neonates with PWS are not always observed at birth but most often during the first days. A normal Apgar score includes normal muscle tone. Cases with both low and normal Apgar scores in PWS have been published previously. Their data call attention to a possible clinical change during the first days. One question to be considered is if any normal neuronal or hormonal stimuli gradually disappear.

FREQUENCY AND RISK FACTORS OF SEVERE SCOLIOSIS IN PWS – Toshiro Nagai -- Japan

They studied 156 patients with PWS - The frequency of scoliosis in Japanese patients was 41 % which was lower than that of the Caucasian populations studied. The frequency of severe scoliosis was about 10 % of all patients and 25% of the patients with scoliosis. With respect to the shape of scoliosis lumbar type was the most common and double-curve type had a tendency of rapid progression. All of these patients with severe scoliosis had surgical indication. About half of the double-curve type showed severe scoliosis indicating that this type should be carefully monitored

EXENATIDE (BYETTA®) INCREASES POSTPRANDIAL FULLNESS WITHOUT SIDE EFFECTS IN PRADER-WILLI SYNDROME - A PILOT STUDY - Alexander Viardot -- Australia.

(Byetta®) is a drug recently developed for the treatment of type 2 diabetes, and it has been demonstrated to have beneficial effects on appetite suppression and weight loss, but it also has significant side effects which limit its use. Eight subjects with PWS and eight obese controls were put on Byetta. The drug significantly increased fullness, but did not reduce appetite, in both groups.

This is the first report on the use of Byetta in PWS that they feel demonstrates that this drug is well tolerated in these subjects. Their observation of suppressed insulin levels and unchanged ghrelin levels challenge previous reports and hypotheses on the mode of action of this drug, suggesting delayed gastric emptying might be an important mechanism (*my comment – it might also be a risk factor for PWS due to the high prevalence of gastroparesis*) which should be assessed in future studies. Larger prospective studies should follow to investigate whether chronic administration of Byetta will lead to decreased food intake and weight loss in PWS, and if it is a safe drug for use in individuals with PWS.

TWO YEARS OF GROWTH HORMONE THERAPY IMPROVES BODY COMPOSITION IN ADULTS WITH PWS – RASMUS SODE-CARLSEN, Sweden

In this first large scale, long-term placebo-controlled study of 46 patients, the improvement in body composition by GH treatment in adults with PWS was confirmed. No side effects were observed.

BRAIN DEVELOPMENTAL DIFFERENCES IN PRADER-WILLI SYNDROME DETECTED BY DIFFUSION TENSOR IMAGING – Ken-ichi Yamada – Yasuhiro Kido -- Japan

The study provides objective evidence that individuals who have PWS indeed have developmental differences in specific areas of the brain. The findings

provide not only new insights into developmental pathophysiology, but also an opportunity for interventional strategies for the behavioral issues in PWS.

NOTES FROM INVITED SPEAKERS PRESENTATIONS:

Daniel J. Driscoll, M.D., Ph.D. FAAP, FACMG -- USA
CLINICAL AND GENETIC OVERVIEW OF PRADER-WILLI SYNDROME

The following are a few of the genetic issues Dr. Driscoll discussed:

PWS is a model system for:

- Obesity and nutrition
- Genomic imprinting and epigenetics
- Endocrinology issues

PWS results from the loss of function of several imprinted genes in the 15q11.2 region. One gene cluster called *HBII-85* (also called *SNORD 116*) seems to be particularly important since two rare patients have been found that only have a small deletion of this gene cluster. These two individuals have many (including early-onset obesity), but not all the clinical features of PWS.

There are two main classes of deletions (Type 1 and Type 2). However, about 10% of deletion patients have a unique deletion size which results in a milder or more severe phenotype depending on the area deleted. Unique deletion subjects will aid scientists in determining the function of the various genes in the PWS region.

Dick F. Swaab, M.D., Ph.D. -- The Netherlands
PRADER-WILLI SYNDROME: GENE EXPRESSION AND PREMATURE ALZHEIMER DISEASE

Now that more persons with PWS are surviving longer, it seems that we find premature aging and dementia, probably of the Alzheimer Disease (AD) type, as a new challenge. Obese patients might be prone to early-onset AD. Also, lack of sex hormones may add to the risk factors.

Special thanks go to Dr. Shaun-Pei Lin of Taiwan for his leadership and dedication to the families in his country dealing with PWS, and his heart for the families of the world.
