Prader-Willi SYNDROME ASSOCIATION (USA)
SAVING AND TRANSFORMING LIVES

Central Adrenal Insufficiency in PWS: Updated for 2018

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Central Adrenal Insufficiency (CAI) was identified as a possible risk in PWS in 2009 by a team of researchers in The Netherlands. At that time, PWSA (USA) recommended testing all individuals with PWS for this potentially life-threatening deficiency. Since 2009, other researchers have published studies which did not find the high levels of deficiency found in the original paper. These studies, done around the world, did not support this and did not find a significant risk of CAI. A review paper, summarizing all such studies, was published in 2016 and it recommends that physicians continue to test for this deficiency, but that CAI may not be common in the PWS population; they noted that it appears less likely in adults. These authors encouraged more research in this area.

Central Adrenal Insufficiency is caused by the lack of pituitary Adrenocorticotrophin Hormone (ACTH) which acts upon the adrenal gland hormones. The adrenal glands make three hormones – cortisol (for energy and sugar balance); androgens (male-like hormones, which stimulate underarm and pubic hair growth) and aldosterone (controls salt balance). The adrenal glands also make adrenaline, which is not affected by ACTH or the pituitary gland.

Cortisol hormone levels vary during the day, with a strong burst before morning, and variations as needed for body stresses. CAI is a very rare condition and a difficult diagnosis to make. The testing is complicated, and the most accurate tests are potentially dangerous or unavailable in the USA. When an individual has a tumor of the pituitary gland or is born without a pituitary gland, the diagnosis is clear; these are the typical reasons for having CAI.

Individuals who take cortisol pills daily must wear medical alert bracelets and be provided with an emergency injection to carry, in case of a serious injury or illness, such as becoming unconscious. There may be some individuals who produce normal daily quantities of cortisol, but who need that “stress dose” whenever they undergo surgery or have a significant illness or injury.

If your child is currently on cortisol, you should continue this medication, and discuss these new findings at your next appointment. It is dangerous to suddenly stop taking cortisol. There are people with PWS who clearly have inadequate cortisol production and who benefit from this medication.

References

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