

**Prader-Willi Syndrome
BRAIN & TISSUE AUTOPSY**

Below is information on autopsy and brain tissue procedures. Please immediately contact the national Prader-Willi Syndrome Association (USA) for assistance and further instruction on the autopsy of an individual with Prader-Willi syndrome. Contact:

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Dick Swaab, M.D., a leading specialist in the world on PWS brain studies advises:

[When performing an autopsy always remember] the most important organ, the brain.

The simplest [way] is to carefully cut the optic nerves and pituitary stalk before lifting the brain, in order not to damage the hypothalamus.

Then remove a piece of cerebellum and put it in the freezer for later genetic investigations. Put the entire brain in buffered formalin (10%, = 4 % formaldehyde), which is the conventional way of fixation all over the world.

After 2 weeks fixation of the brain is enough for transportation by overnight mail, if possible, to Amsterdam. We can arrange the transport and will give the PWS researchers the possibility to do serious research on the material.

Of course, more sophisticated techniques (dissecting of well defined structures and freezing of samples) can be discussed with specialized labs.

Dr. Merlin Butler states, "It is important to get some fat and muscle biopsies (visceral & peripheral) from the stomach and thigh, and samples from the liver and pancreas. This adrenal gland is at question, so size and sample of this gland would be helpful."

Dr. Dan Driscoll adds, "Freeze a piece of cerebellum and some blood in a purple top so that there is material to make a genetic diagnosis since formalin fixed tissue is not good for DNA diagnosis. Pack vial of blood in crushed ice.

Request blood for DNA on all brain collections.

NOTE: The PURPLE TOP blood tubes kept cold (frozen or refrigerator temperature) are best for DNA collection, and blood for DNA should be requested on all future brain collections. "