

**PROTOCOL FOR PROTEIN LOAD TEST FOR THE DIAGNOSIS OF
HETEROZYGOTES FOR ORNITHINE CARBAMOYL TRANSFERASE (OCT)
DEFICIENCY**

This protocol has been designed to test for carriers for OCT deficiency in two ways:

- Firstly by collecting a fasting sample of urine (**FASTING SAMPLE**) for analysis of orotic acid/creatinine and comparison with our control population.
- Secondly by collecting three timed urine samples over the 4 hours of the test. **SAMPLE 1** is collected immediately prior to eating the cooked chicken, **SAMPLE 2** collected during 0 to 2 hours after the meal and **SAMPLE 3** collected during 2 to 4 hours after the meal. Analyses of orotic acid/creatinine on these samples are used to calculate the ratio of orotic acid excretion from 2 - 4 hrs over 0 - 2 hrs after the meal of protein. The absolute results and the ratio are compared with the ratios found in our control population (see **Journal of Inherited Metabolic Disease Vol 24, 1 pp 5- 14**).

It is ABSOLUTELY essential that the instructions for collection of urine are followed exactly.

Preparation of the Meal

- A. Height and weight of patient need to be supplied to the clinician or dietitian well before the test commences.
- B. A meal of steamed chicken breast (no fat, no skin) is to be prepared and cooked. Raw chicken breast contains 22.6 g protein/100 g wet weight. The raw chicken should be weighed to give a calculated load of 1.0 g protein/kg body weight, if >40 Kg and <75 Kg. Give 35 g protein/M² surface area if body weight is <40 Kg or > 75 Kg.

Preparation of/by the Patient

- A. Have nothing to eat or drink, except for water, from 10.00 pm. on the night before the test. Medications may interfere with the test. Any medications that are absolutely necessary must be discussed with the laboratory [Dr. Wilcken (02) 9845.3650; Dr. Carpenter (02) 9845.3123, Ms Green (02) 9845.3133].

The collections of urine start on getting up in the morning and continue until the end of the 4 hour test. All of each specimen passed must be collected, put into a clean container and labelled with name, **time** and date.

- B. Collect the first urine passed after getting up, put all of it into a clean container and label with name, **time** and date (**FASTING SAMPLE**). The specimen should be kept cool. If at all possible, please refrain from passing urine again until the start of the test. If that is not possible, collect again into a separate clean container and label as before with the **time**, etc. . A glass of water (250 mL) may be drunk.
- C. On arrival at the appointed place, you will be asked to empty your bladder again (**SAMPLE 1**) and then immediately to start eating a meal of chicken and finish it within 25 minutes. It is helpful to wash down the meal with a glass of water (250 mL). Ideally urine should be passed at exactly 2 hr (**SAMPLE 2**) and (**SAMPLE 3**) 4 hr after the meal. A glass of water (250 mL) may be drunk during this period.

N.S.W. BIOCHEMICAL GENETICS SERVICE
THE CHILDREN'S HOSPITAL AT WESTMEAD

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DATA SHEET FOR PROTEIN LOAD TEST

DATE OF TEST:

NAME:

DATE OF BIRTH:

HEIGHT (cm): WEIGHT (Kg):

SURFACE AREA (m²): (if necessary)

CALCULATED PROTEIN LOAD:(raw chicken)
(1.0g protein/kg BODY WEIGHT if >40 Kg and <75 Kg;
35g protein/m² SURFACE AREA if <40 Kg and >75 Kg)

ANY REACTION TO TEST:(nausea, headache, etc.)

IMPORTANT

- It is essential to collect all of each specimen of urine passed.
- It is essential to record the time and total volume of each collection.
- It is essential to empty the bladder immediately before starting to eat.

PROCEDURE

TIME

TOTAL VOLUME

URINE COLLECTION BEFORE THE MEAL

FASTING SAMPLE First urine after rising this morning	_____	_____
Other urine (if any) before appointment	_____	_____
SAMPLE 1 At the start of the meal	_____	_____ ****

TAKING THE MEAL (TO BE EATEN WITHIN 25 MINUTES)

Start of eating	_____	Same as ****
Finish of eating	_____	No sample

NOTE: If all of the chicken is not eaten then Weigh the Cooked Chicken not eaten.....g and CONTACT THE METABOLIC DIETITIAN

URINE COLLECTION AFTER START OF MEAL

SAMPLE 2 Between 0 - 2 hours (if any)	_____	_____
SAMPLE 2 Bladder emptied at 2 hours after start	_____	_____
SAMPLE 3 Between 2 - 4 hours (if any)	_____	_____
SAMPLE 3 Bladder emptied at 4 hours after start	_____	_____

NOTE FOR LABORATORY SUPERVISING THE TEST: SAMPLES for Biochemical Genetics

After the total volume of each sample has been thoroughly mixed and carefully measured, approx. 20-30 mL may be transferred to a small screw-cap container, labelled clearly with name, date, time, total volume and frozen. Please keep the samples frozen during transport to Biochemical Genetics.