Newborn Screening

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JOANNA AND JAMES: BABIES AT RISK RESCUED BY NEWBORN SCREENING

by Fedelynn "Chat" M. Jemena

Most of us believe that the only risks a baby faces are any or a combination of the following: starvation, abuse, kidnapping, or inept caregivers. All are nerve-racking situations stemming from the environment. But there are dangers which, inspite of the most careful of parents, still manage to put a baby's life in a balance. They may come sneaking in like proverbial thieves in the night. Such has happened to babies Joanna Paula and James Michael.

Joanna is a lively, doughnut-loving, wide-eyed, six-month old who suffers from Congenital Adrenal Hyperplasia (CAH), a genetic disorder which occurs in one out of every 12,000 births. Literature describes it as a flaw in the pathway that produces some hormones causing the baby to lose salt via the urine and to have high levels of male hormones.

Joanna was born coal-black but her parents, Romulo and Teresita, accepted it because Romulo had leathery brown skin. Her clitoris was elongated and everyone mistook it for a penis. Her parents even had her registered as John Paul. Luckily, newborn screening was done on her third day of life. It involved getting a sample of blood through heelprick method. On the 10th day of John Paul, an urgent call from Australia came informing the Secretariat office that Joanna's sample showed a significantly elevated result for CAH. Chinese General Hospital was immediately notified and they in turn tried to contact the family through telegrams and home visits. These measures seemed futile since the given address was confusing. Fortunately, a hospital personnel helped the doctors locate her. She was admitted just before severe complications could develop.

Joanna's parents initially could not believe that their cute little baby had CAH. "*Sa umpisa, medyo masakit, pero tinanggap na rin namin*," Teresita explained. Romulo, on the other hand, was the typical forebearing Filipino: "*Eh, wala na rin namang magagawa.*

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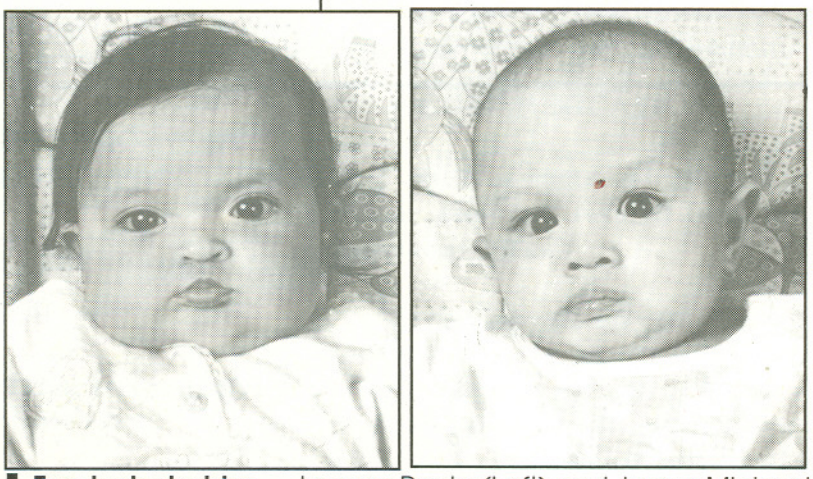

Two lucky babies ... Joanna Paula (Left) and James Michael (Right).

PROJECT UPDATE

For the past 7 months, 13 367 newborns have been screened at the 24 participating hospitals of the Philippine Newborn Screening Project. Of the 39 newborns who yielded positive screens, 5 have been confirmed - 3 with congenital hypothyroidism (1:4 456) and 2 with congenital adrenal hyperplasia (1: 6 683).

The Secretariat Office is currently monitoring all babies with positive screens and positive results. Home visits are being conducted for patients with positive screens and who have been lost to follow up. False positives and false negatives must be reported immediately to the office. The algorithms for follow up and management must be strictly followed. All these will help us validate any future recommendations of the group.

The quality of the filter paper cards being submitted have improved bringing the reject rate to almost zero. Congratulations to all the residents involved in the project.

We commend the coordinators of the hospitals who are working hard to improve the performance rate of their hospitals. Every hospital's performance is being monitored by the office. In the last meeting of the overall coordinators,

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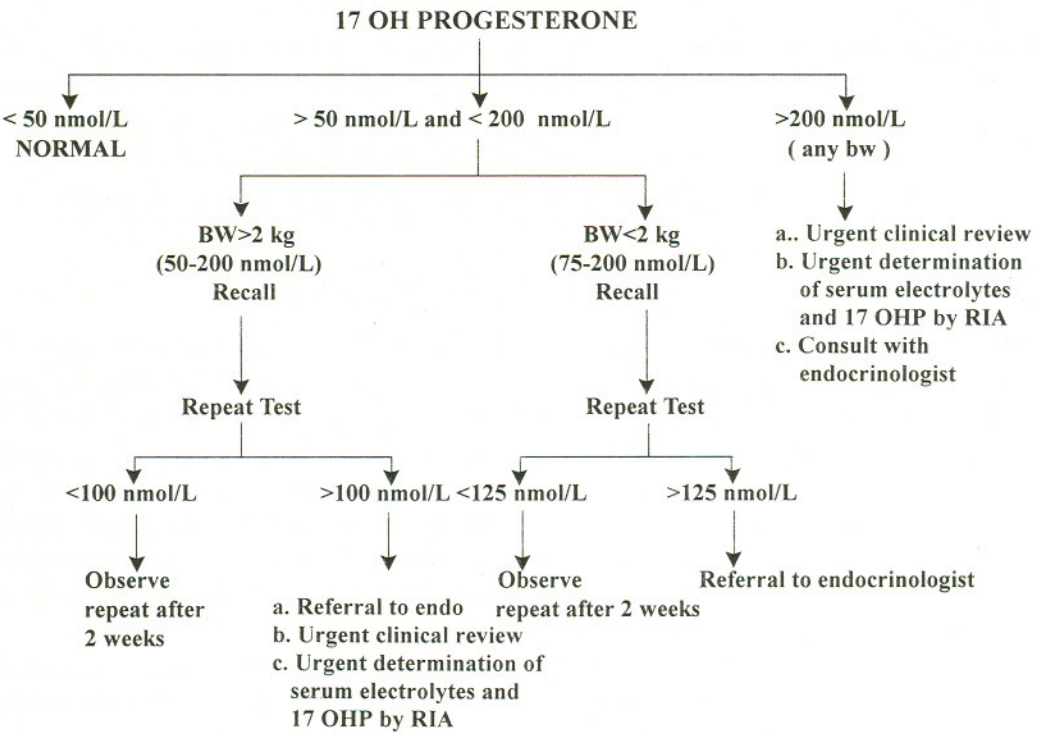
NEWBORN SCREENING
FOR CONGENITAL ADRENAL HYPERPLASIA

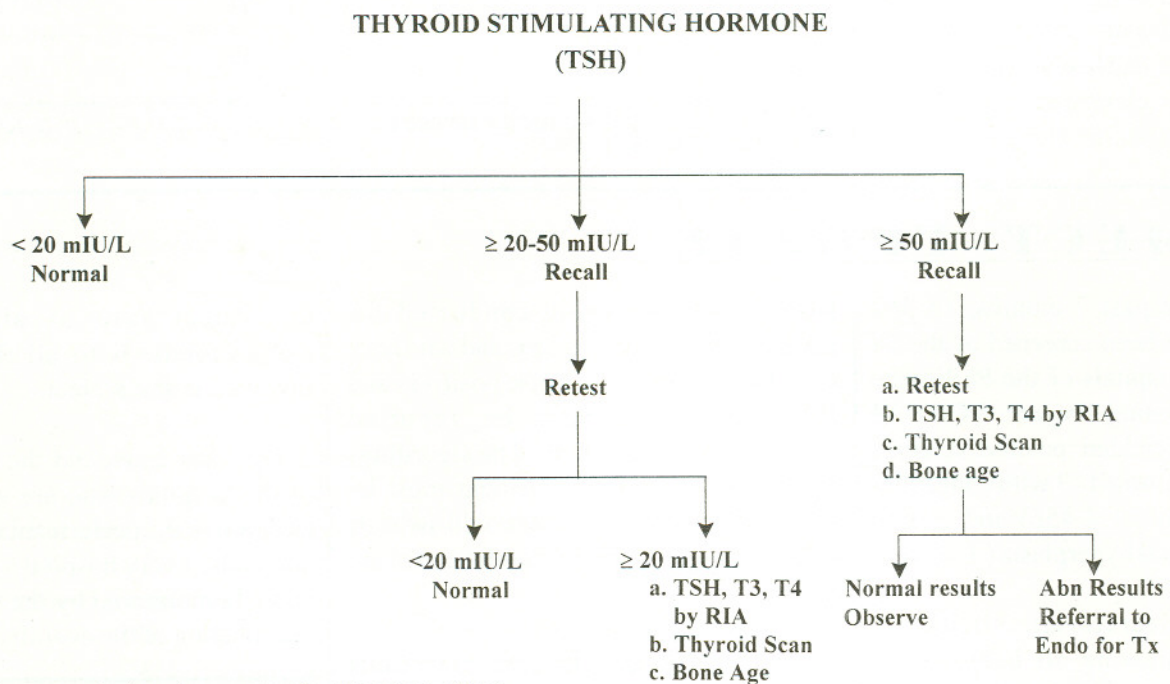
Guidelines for Action and Confirmatory Testing*



as agreed upon during the coordinators meeting; proceed to **Confirmatory Testing if patient is manifesting signs and symptoms of CAH while waiting for the screening result*

NEWBORN SCREENING
FOR CONGENITAL HYPOTHYROIDISM

Guidelines for Action and Confirmatory Testing*



** as agreed upon during the coordinators meeting*

QUESTIONS & ANSWERS

SAMPLE COLLECTION

How do we get good samples?

A very successful method for getting good samples is to make two quick successive pricks on any spot found on the lateral side of a warm heel. Of course, not everyone gets it right at the first try but practice makes perfect.

Do you advise the blood extraction method in getting a sample?

No. The heel prick method is still preferable. However, if the baby will be having a blood extraction for other laboratory tests, then, you may.

Why are our samples rejected?

** It could be any of these reasons:*

- *contamination*
- *inadequate blood sample*
- *insufficient drying*
- *layering*
- *sample is less than 48 hrs.*

(Please refer to the PNSP manual.)*

Can we still screen babies who are more than one month old?

Yes, but it defeats the purpose of the study which is the early screening of newborns for early diagnosis and treatment of inborn metabolic disorders.

RESULTS

Does a positive screen mean a positive result?

No. All patients with a positive screen must be re-tested Follow the guidelines for action and confirmatory testing

How soon do we receive the results of the screening from Australia?

For babies with positive screens, we are informed the day the samples are run in Australia. This is approximately 4-7 days from the date we send the package to Australia. Soon after the results are received by our Secretariat Office, we immediately fax these to the participating hospital concerned and inform the Overall Coordinator.

For babies with normal screens, results are sent by batches every two weeks by fax. We send the results through mail and through fax and addressed to the Overall Coordinators to make sure that they are received.

Some of our results are missing. Why is it so?

All the samples we receive are always sent to Australia on the same day. Only contaminated samples are not. In some cases, patients could not be located because the patient's names and names of hospitals were misspelled which is a result of illegible handwriting. Because of this, patients are logged in the wrong hospitals. Please write legibly and do not abbreviate the hospitals names.

SUPPLIES

How often do we get our filter papers?

We originally issued filter papers that would last 3 months. Unfortunately, because of wastage, we ran short of supplies. Currently, we only issue filter papers upon demand.

How soon do we get our supplies?

We make it a point to send out supplies like filter papers, receipts, consent forms etc., on the very day we receive the request. The package usually arrives the afternoon after it is sent from our office. Because it takes at least a day of waiting, please request for your supplies at least a week, before supplies run out to prevent any lapses in sample collection. We always address these packages to your Overall Coordinator.

FEDEX PICKUP

Who should we call regarding pickup of our samples?

Please contact Mr. Ed Barrios at the Fed Ex office before 3:00 p.m. at telephone number 8913595. If you have any complaints regarding pickup, please let our Office know so we can personally address the problem to Fed Ex. We are very much concerned about this because another reason for the delayed results is the poor pickup of samples.

COMMUNICATION

Who receives communication (i.e. memos, supplies, results) from the Secretariat Office?

*It is the responsibility of the **Overall Coordinator** to relay relevant information to the consultants, residents and nursery staff. We recommend a regular dialogue with the Nursery Staff to improve the performance of the project.*

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... Joanna (continued from page 1)

Pinagkaloob sa amin na ng Diyos kaya tinanggap na rin namin." The dedicated doctors of Chinese General helped them in understanding and coping with the condition.

Thus, John Paul became Joanna Paula because chromosomal tests proved the baby to be a girl. Her tests also revealed that she would need imported medications such as Fluorinef and Hydrocortisone for the rest of her life. As the family is poor, the doctors are helping in obtaining the medication from abroad. The Joanna of today would not strike you as a baby who would die without medications. You would not even think of her as the same baby as she was because she looks healthy and had shed her coal-black coloring. Her hormones are managed well and her penile-like clitoris is decreasing in size. However, she would still need an operation to correct this condition.

Her parents are grateful that Joanna has survived when other children of her age and condition had already died. Their priority concerns now are the procurement of medication and the changing of their daughter's records in the City Hall Registry.

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Venerando and Leilani were also careful parents. They wanted their second child to be as healthy and as good looking as their eldest, a boy. James Michael is certainly goodlooking, a mestizo and a spitting image of his father. But he, too, had a condition that could have resulted in mental retardation had it not been detected and treated early. He had Congenital Hypothyroidism (CH).

Congenital Hypothyroidism is a rare condition that occurs in one out of every 4000 births. With this disorder, the thyroid hormone, which is essential to the growth of the brain and the body, is either low or absent.

Fortunately, they were approached by an NBS advocate at the United Doctors' Medical Center three days after James's birth. Leilani recalled that their attitude at that time was,"*Sige, pa-screen na; wala namang mawawala. Buti iyon, habang maaga, malaman na namin kung may sakit siya.*" Leilani was the only one at home at the time she received the call, saying, "*When I received the call that our child appeared positive for one of the illnesses, medyo nakakalula. Bakit ganoon. Ingat-ingat pa naman ako noong nagbubuntis pa ako. Our first child is OK. Sabi ng nakausap ko, ipa-retest ko for the second time para malaman kung positive nga, para maagapan sa therapy. The next day, pumunta kaagad kaming magpamilya.*"

Venerando and Leilani are coping well despite their worries.. Their hope now is for all the babies born in participating hospitals to be screened. They are hoping that the Government and the Private Sector would support the program and every baby born in the country will be screened including those born in their respective homes.

STUDY UPDATE

June 1996 to January 31, 1997

Total number of Patients entered in the Study:	**14, 204**
Accepted samples:	**13, 367**
Rejected samples in Manila:	**837**
Rejected samples in Australia:	**92**
Total No. of Patients Screened:	**13, 367**
Total No. of Patients with Positive Screening:	**39 (1:343)**
Breakdown of Patients:	
CAH	**9**
CH	**24**
PKU	**1**
TYR	**1**
GAL	**1**
With multiple elevations	**3**
Total No. of Patients with Confirmed Results:	**5**
Breakdown of Confirmed Patients:	
CH	**3 (1:4,456)**
CAH	**2 (1:6,683)**

... Project (continued from page 1)

the group agreed to aim for at least 50% of newborns to be screened in every hospital. The graph below presents the overall performance of the project. As of January 1997, we screened 39% of newborns in the 24 hospitals. We are all eagerly looking forward to the day when all newborns in the participating hospitals will be routinely screened.

Overall Percentage of Newborns Screened among the 24 Hospitals