CONCLUSION

- Though metabolic disorders are rare, one needs to suspect a metabolic disorder while working with the disabled.
- Cases diagnosed to have metabolic disorders early in life, could have been prevented from being disabled and even death avoided, if appropriate steps had been taken.
- Adhering to the special diet is imperative, as the wrong food can cause multiple disabilities and even death.
- Insufficiency in bodily metabolites' is seen through TMS testing due to AEDs, deficiencies similar to vitamin deficiencies thus causing abnormal meta bolic functioning by itself leading to disability.
- Appropriate supportive therapy need to be followed when children are on AEDs.

OUTCOME MEASURES:

- The clients with metabolic disorders will be advised appropriate diet, supportive supplements for their condition and also on methods to improve skills in them, through the Early Intervention services provided.
- For clients unable to afford dietary supplements, formulae feeds were secured on grounds of compassion (partner organizations).

RECOMMENDATIONS

- Reduce or exemplify duty tax on these dietary foods imported from abroad and produce these modified diets indigenously.
- Collaborations with various institutes of inter-disciplinary nature towards the betterment of the Multiply Disabled.

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RESEARCH at National Institute for Empowerment of Persons with Multiple Disabilities

BIOCHEMICAL PROFILING OF PERSONS WITH MULTIPLE DISABILITIES

Status: Completed (2010 – 2012)

Abbreviations: Physical Medicine & Rehabilitation (PMR), Antiepileptic Drugs (AEDs), Liver Function Tests (LFT), Cerebral Palsy (CP), Mental Retardation (MR), Developmental Delays (DD)

Abstract

Inborn Errors of Metabolism (IEMs) are biochemical disorders that can result in multiple disabilities. If timely intervention is given in the form of formulae diets, enzyme replacement therapy, supportive therapy and early intervention, the resultant multiple disabilities can be avoided. One such diagnostic study was initiated by the National Institute for Empowerment of Persons with Multiple Disabilities, which not only diagnosed the cause of the disabilities, but also instituted therapy in form of diets and medication changes. The study showed IEMs existed in clients seen at NIEPMD and caused multiple disabilities. While some had already become disabled, the progress and severity of the condition has been lessened through diets, medication changes and early intervention.

Introduction:

IEMs are enzyme defects in the pathways of metabolism (Garrod, 1909). They have a general prevalence of 1 in 800 to 1 in 2500 live births (Ananth, 2009), causing developmental delays leading to Multiple Disabilities. By timely diagnosis and instituting appropriate therapy (Talkad, 2006), the deterioration associated with the disorder can be minimized and even normalcy in life can be achieved.

Aims & Objectives:

The Department of PMR initiated a diagnostic study to find out the underlying causes of Multiple Disabilities. IEMs as a causative factor are being considered in this diagnostic study.

Methodology:

- Sample size of 25 clients were selected, those who had dysmorphic features / markers of biochemical abnormalities.
- A drop of blood was spotted on to a filter paper and sent the very same day to NIMHANS, Bangalore. The sample was tested through Tandem Mass Spectrometry (TMS) at the Neurochemistry dept. which is headed by Dr. Rita Christopher.
- Routine biochemical screening tests were done at a Muthukumaran Medical College, Chennai. Clients were admitted overnight and blood samples were drawn accordingly. The Biochemistry dept. is headed by Dr. Renuka Raveendaran who foresaw the testing and analysis.
- Disorder specific tests, samples of blood/urine were sent to Institute of Human Genetics, Ahmadabad. The lab is managed by Dr. Frenny and Dr. Sheth.

Inclusion criteria

Clients with multiple disabilities registered at NIEPMD.
Clients with markers / suspicion towards IEMs.

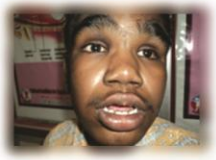
Metabolic markers / suspicion (list is not exhaustive, source www.metagene.de)

1. Low Birth Weight
2. APGAR score
3. Delayed Birth Cry
4. Hypothyroidism
5. Microcephaly
6. Macrocephaly
7. Cataracts
8. Brittle hair/ Blonde hair
9. Low set ears
10. Hypertrichosis
11. Hypertelorism
12. High arched palate
13. Tongue thrusting
14. Hypertonia
15. Hypotonia
16. Syndactyly
17. Urine / Body odour unusual
18. Psychiatric problems

Exclusion criteria

Confounding factors, such as Sepsis, TORCH, Birth Asphyxia etc.

Case 1



MUCOPOLYSACCHARIDOSIS

Symptoms

Course Facial Features
Mental Retardatio
Vision Problems

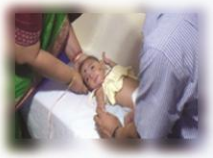
Case 2

MAPLE SYRUP URINE DISEASE

Sysmptoms

Unusual Urine odor
Unusual body odor
Seizures
Failure to thrive
Head lag

Case 3



PROPIONIC ACIDEMIA

Symptoms

Blonde hair
Sparse hair
Failure to thrive
Seizure

Case 4

LOWES SYNDROME

Sysmptoms

Mental Retardation
Cataract Operated
Renal Problems

Case 5

DRUG INDUCED (AEDS)

Sysmptoms

Head lag
Drowsy
Seizures

Case 6

DRUG INDUCED (AEDS)

Sysmptoms

Microcephaly
Lethargy
Seizures
Frequent illness

Case 7

HOMOCYSTINURIA PREVENTED

Sysmptoms

Skin color changes
Lens Dislocation

Case 8

HOMOCYSTINURIA

Sysmptoms

Hypotonia
Lens Dislocation
Thin long hands, fingers
Skin color changes

Case 9

HYPERLIPOPROTEINEMIA

Sysmptoms

Low vision
Spastic
MR
CP

Case 10

MUSCLE MYOPATHIES

Sysmptoms

Developmental Delay
Vision Impairment
MR

Case 11

TRANSIENT
HYPERPHOSPHATESEMIA

Sysmptoms

DD,CP,MR, LV ,
Elevate LFT &
Alkaline Phosphatase