Neuroimaging in Mentally Retarded Iraqi Children

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Muna A.Gh. Z

FICMS.(rad), Lecturer, Department of Surgery, Al-Mustansiriya College of Medicine

Abstract:

Background: mental retardation is a common clinical problem in pediatrics, it present during infancy or preschool years as developmental delay.

Objectives: to determine the diagnostic yield of neuroimaging in children with mental retardation of unknown origin.

Methods: Neuroimaging (Computerized Tomography CT scan) was performed in a total of 59 patients with developmental delay/mental retardation, in AL-Yarmuk Teaching hospital in Baghdad from September 2006 to June 2008 where no etiological diagnosis could be made following clinical examination and preliminary investigations.

Results: Forty one (69%) had abnormal neuroimaging findings of which 13 (22%) were specific abnormalities useful in arriving at etiological diagnosis. Positive outcome of neuroimaging increased with the severity of mental retardation, presence of microcephaly and other neurologic deficits other than MR.

Conclusions: Neuroimaging (CT scan) should be the standard clinical practice for a child with global developmental delay where no cause is apparent after examination and relevant investigations.

Keywords: Neuroimaging, Mental, Retardation

Introduction:

Mental retardation refers to incomplete or insufficient general development of mental capacity with sub-average intellectual functioning (IQ <70) existing concurrently with related limitation in two or more of the following applicable adaptive skill areas, Communication, self care, home living, social skill, community, self direction, health and safety, Functional academics, leisure and work. It is a common clinical problem in pediatrics with on estimated prevalence of 3% and it most often presents during in -fancy or pre-school years as developmental delay [1,2,3]. The male-female ratio is 1.6:1 in mild retardation, but close to unity in severe retardation [4].

Early identification of affected children, is important so that they may benefit from early intervention programmes. An additional goal in the evaluation of developmental delay in children, is the determination of a specific cause for the disability. This can provide information regarding the possible pathogenesis, prognosis, recurrence risk, prevention of recurrence and specific medical intervention as well as the critical question most of them posed by the families of these children. Several clinical series suggest that a definite cause for mental retardation can be identified in 40-60% of all patients undergoing evaluation [5,6,7,8].

In the remaining patients the cause remain obscured, even after extensive clinical work up and laboratory studies. By definition, an individual with mental retardation has a functionally abnormal brain, which may or may not have a developmental basis in morphogenesis or histogenisis. Structural brain abnormalities has been demonstrates in 34-98% diseased severely retarded patients undergoing complete neuro pathological studies[9,10,11]. Fewer numbers of living patients (9-60)% are reported to have an abnormality on neuro imaging[12,13]. In the back drop of an anticipated high yield of newer, improved neuro imaging techniques, we conducted this study to determine the diagnostic yield of CT in a cohort of children with developmental retardation of un known origin.

Patients and Methods:

Brain neuroimaging (CT scan), was done in 59 developmentally retarded children where no etiological diagnosis could be made. All patients referred to inpatient radiological department of

Al Yarmuk General hospital in Baghdad for development delay or mental retardation, where the developmental function was less than 70% of the chronological age, the decision to perform CT evaluation, was based on the clinical judgment of the referring pediatrician.

Cases with known history of CNS malformation, like meningitis , encephalitis and obvious malformation like hydrocephalus, metabolic disorders like muco polysacchridoses and recognized syndromes including chromosomal disorders ,known to cause development retardation were excluded from the study, as our aim was to evaluate the role of neuro imaging in providing etiologic diagnosis in cases presenting as mental retardation, but where clinical examination and relevant tests are unsuccessful in providing etiologic diagnosis.

Examination technique: Utilizing the spiral CT scan, somatom plus 4 model 1999 , k.v 140, MAS 120 , minute 39 sec.

Patient Positing: In supine position, with a section plane at 10-25 Raids base line. It is a comfortable position, and reduces lens radiation of the eye.

Head position: Supine-head first, for brain, slick thickness 5mm, for base of skull ,3mm, serial slices from below upward were undertaken.

Result:

Brain CT scan was done in 59 patients with MR, where no etiological diagnosis could be made, following clinical examination and relevant tests. They were 39 male and 20 female. Their ages ranged from 8 months--- 13 years. The bulk of cases fall in the age group (1-3) years. Patients were divided in to two groups:

1-Retarded only: MR without any other neurological deficit (21/59= 35.5%).

2 -Retarded neurologic: MR associated with neurologic features (38/59=64.4%) eg. Seizures, hypotonia, hypertonia, abnormal reflexes etc.

In the 2nd group, the frequency of abnormal neuroimaging was higher. Forty one (69.49%) retarded children had abnormal finding, of which 13 (22%) were specific.

The remaining 28 had isolated atrophy as positive finding of nonspecific significance as it did not provide any diagnostic clue to the etiology. Though the number of cases in different groups is small, our data shows that the percentage of abnormal neuroimaging finding increased with the severity of MR. Table 1 demonstrate findings in both male and female. Where cerebral atrophy was the commonest finding, male \ female (21:7) = (3:1). Male=35.5% female 11.8%. Next to, is a CT scan image were no findings seen , male=10 =16.9%. Female=8 =13.5%. Five cases of microcephally did not show abnormal CT findings.

Table 1 Distribution of the study sample according to CT finding in both males and females.

CT findings (cerebral)	***Males***		***Females***	
	No	***%***	***No***	***%***
Atrophy	21	35.5	7	11.8
No findings	10	16.9	8	13.5
Infarction	3	5.1	2	3.3
Demyelination	2	3.3	1	1.7
Periventricular calcifications	2	3.3	1	1.7
Pachygyra	1	1.7	0	-
Holoprocencephaly	0	-	1	1.7

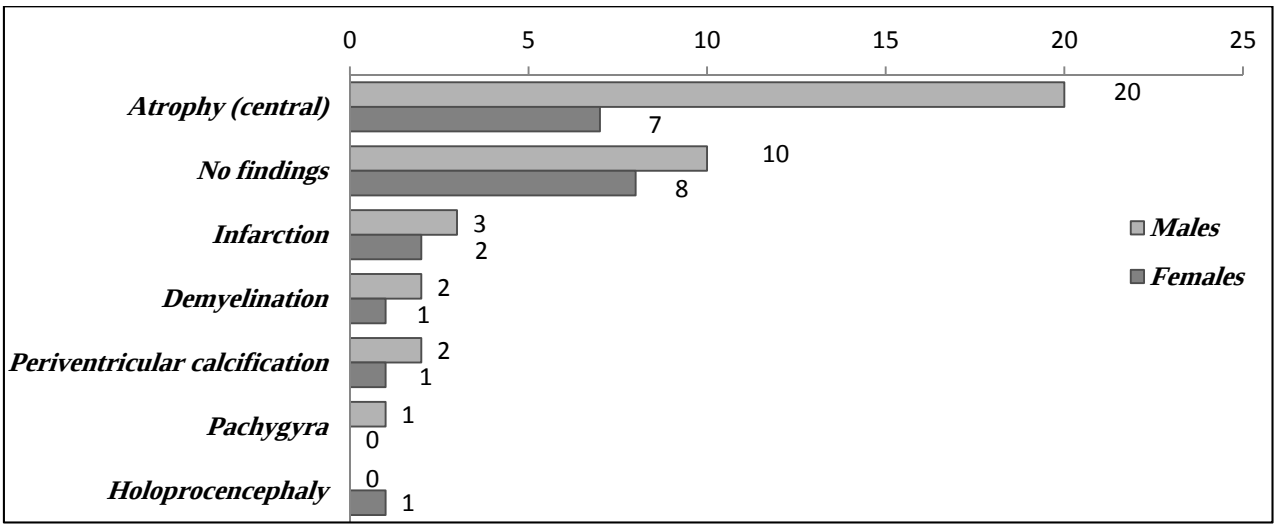

Figure 1: The distribution of the study sample according to the CT findings in both males and females.

White matter abnormalities were seen in three cases, in one case, the finding was suggestive of demyelination with evidence of leukodystrophy, the patient had profound developmental delay and marked hypertonia.

In the other two cases, findings are nonspecific for metabolic disorders, their presence, and guides farther direction of investigations to give definite diagnosis. Though all possible efforts were made to exclude cases of antenatal infective insults or known perinatal asphyxia, in 8 cases, imaging finding were in favor of some acquired brain damage. In five cases , cerebral infarction raise the possibility of perinatal asphyxia , while in two cases , per ventricular calcification suggest congenital infection .Congenital Morphologic abnormalities identified in two patients as a cause of MR, one patient with pachygyra, did not have focal neurological deficit, but a smaller than normal head circumference, one patient with holoprosencephaly had spasticity and seizures.

In one case of per ventricular calcification, subependymal tumors seen on CT scan, suggestive of Tuberous sclerosis. The clinical diagnosis was not obvious as the patient lack seizures or characteristic skin lesions.

Table 2: Distribution of study sample according to the type of atrophic changes.

Atrophy	***Males***		***Females***	
	No	***%***	***No***	***%***
Fronto-temporal	21	35.5	7	11.8
Generalized	10	16.9	8	13.5
Semi-lobar	3	5.1	2	3.3
Frontal	2	3.3	1	1.7
Temporal	2	3.3	1	1.7

Two cases of generalized atrophy were associated with microcephaly. Both generalized and fronto-temporal atrophy were common.

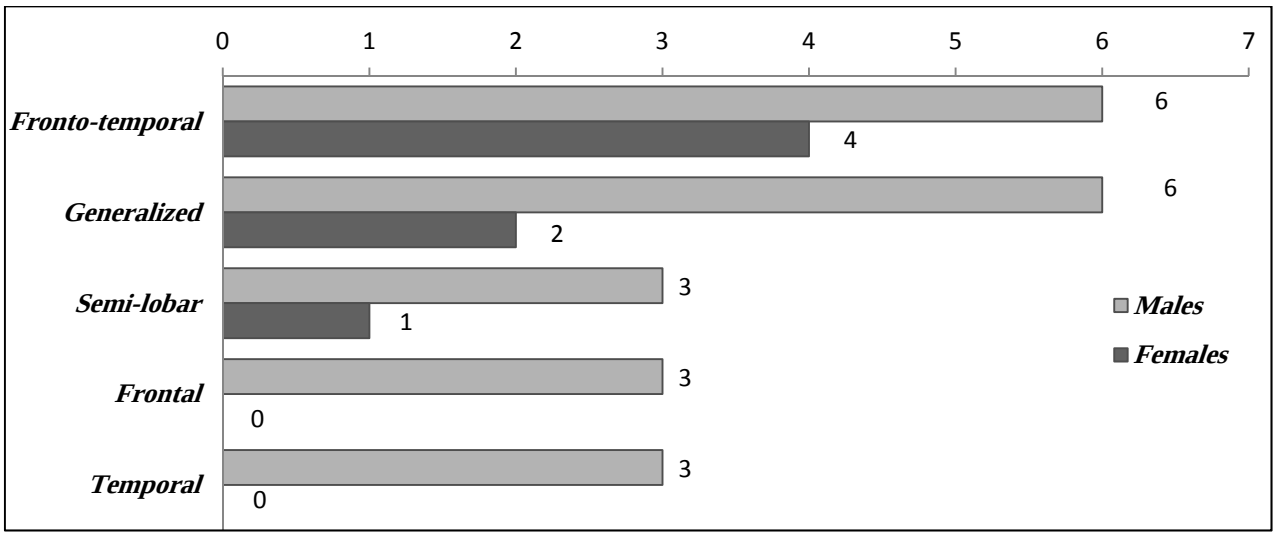

Fig2: The type of atrophic changes.

Discussion:

The aim of investigating a child with MR or developmental delay is to reach etiologic diagnosis. Though not always diagnostic, neuroimaging provides useful diagnostic information in a relatively high percentage of retarded children, where no other diagnostic clue is available. The sensitivity of neuroimaging in this cohort with a 22% rate of positivity exceeds that of other laboratory tests. These findings are comparable to those of kjos et al who reported a 34% positivity of MRI in idiopathic developmental retardation[(12)]. This can be attributed to the versatility of imaging in providing diagnostic information about a variety of condition associated with abnormal morphology, structural malformations, metabolic disorders, vascular or infective insults. In the evaluation of 7 cases of microcephaly, only two had abnormal findings (28.5%) it is lower than those reported by Steinin et al and pander A. etal which revealed an abnormality of 67% and 56.25% of cases respectively. [(14, 15)]

When MR was associated with other additional neurological deficits, the likelihood of positive imaging findings suggestive of specific etiologic diagnosis increased to 64.4%. These findings are similar to Pandy A. et al which revealed 65% of cases. This group is especially baffling to clinicians seeking to establish a definite cause of MR. congenital morphogenetic abnormalities represent developmental disorders, occurring early in gestation. In the present study, one patient had abnormal migration defects namely pachygyra and in another, Cleavage defects, these have various etiologies like monogenic, chromosomal or even infective causes and thus, further, investigation are needed. The other important causative factor for mental retardation, which can be diagnosed by imaging, is the group of metabolic diseases. Most of these are associated with demyelination. The clinical diagnosis is very difficult, moreover, due to lack of easy availability of the required battery of tests. Many are missed or at times a metabolic cause is ever not suspected [(16)]. In the present study too, serial cases had imaging findings suggestive of leukodystrophy or other metabolic disorders but none of them gave a clinical clue like history of regression of mile stones of progressive nature, associated features like abnormality of retina, Hepatosplenomegaly which could suggest the possibility of the same. Thus neuroimaging can provide a diagnosis or a diagnostic clue to metabolic diseases. Though very nonspecific generalized cerebral atrophy is also a feature of some metabolic disorders and its presence warrants a proper work up for a metabolic cause.

Prenatal infection is an important cause of brain damage but because of an extreme variability of presentation, clinical diagnosis is difficult. Serological investigations to rule out infections are inconclusive unless they are done in neonatal period or early infancy. Based on history alone, diagnosis of perinatal insult as a cause of brain damage is many times erroneous but,

documentation of imaging findings, suggestive of hypoxia; ischemia or infection is important diagnostic evidence. Though imaging may not provide the exact time of the insult or the specific cause for the same, the parents can be assured of a negligible risk of recurrence.

The application of neuroimaging does not facilitate any specific management in the retarded children, nor does it alter the patient's developmental status. Nevertheless, it is immensely useful in providing valuable information regarding an etiological basis for the retardation. This greatly helps the clinician in counseling the concerned families and it enables the couple to make informed decisions regarding reproductive choices.

In addition an etiologic diagnosis provides for a more accurate prognosis. In the present study, after neuroimaging, the positive results help the families in finding some solace that there was some physical factor in the brain beyond their control that accounted for this lifelong problem. Finding causes help them to accept the problem better and stop their efforts for diagnosis and pay more attention to training and other - (Magnetic Resonance Imaging -MRI) allows a more accurate analysis of cerebral anatomy, better evaluation of myelination and delineation of grey matter-white matter in vivo than CT, yet in this cohort, CT was used rather than MRI because of difficult facilities in providing general anesthesia for children. However, sufficient information's may be achieved by CT to warrant, its use in situations with limited access[(7)].

Conclusions:

It is recommended that if an etiologic diagnosis is not readily apparent after a detailed history, examination and initial investigations, neuroimaging should be the standard clinical practice for a child with global developmental delay. However, it is emphasized that imaging is not a substitute to careful clinical evaluation. Neuroimaging, should be considered in patients without a known diagnosis especially in the presence of micro or macrocephaly and additional neurological findings like hypertonic seizures etc. this concurs with the opinion of consensus conference about evaluation of mental retardation [(13)].

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