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ORIGINAL ARTICLE

Clinical Aspects of Microcephaly Patients Regarding Endocrine Profile

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ABSTRACT

Introduction: Microcephaly is characterized as an occipitofrontal head circumference (OFC) underneath the third centile or more than 2 standard deviations (SD) below the mean for sex, age, and ethnicity. The term 'severe' microcephaly is connected to an OFC more than 3SD below the mean. Microcephaly is associated with a reduction in brain volume and frequently intellectual and/or motor inabilities. The pathogenesis of microcephaly is heterogeneous, extending from hereditary causes to environmental components that can have an effect on developmental process that impact brain size.

Objective: The main objective of this study was to compare the BMI and endocrine profile of patients having microcephaly with age matched to normal siblings in their families.

Materials and Methods:

Study design: Quantitative cross sectional

Settings: Services Hospital Lahore

Duration: 01 year i.e. 1st January 2020 to 30th December 2020

Methodology: This is a quantitative cross sectional study arrangement based on 12 persons. On the basis of microcephaly, the subjects were separated into the two groups: Group I: Subjects with microcephaly (n=10), Group II: Normal kin as controls (n=2). Five families including add up to of 12 individuals was selected. Ten people with microcephaly (cases) and two normal kin (without microcephaly) were taken as controls. Cooperation of the subjects in this study was selected voluntarily and written informed consent was taken to take part in the study from each individual and from their guardians. The individuals and their guardians were educated about the potential benefits and risks of this study.

Results: There was little difference within the mean age of microcephaly individuals as compared to healthy siblings.

Conclusion: In light of this study it can be recommended that there's a significant affiliation of BMI and microcephaly but the affiliation of Leptin, Cortisol, GH and TSH with microcephaly seem not be found as proposed by non-significant results. This may moreover emphasize on heredity perspective of this condition.

Keywords: Microcephaly, Leptin, Growth Hormone, BMI, TSH

INTRODUCTION

Microcephaly is characterized as an occipitofrontal head circumference (OFC) underneath the third centile or more than 2 standard deviations (SD) below the mean for sex, age, and ethnicity. The term 'severe' microcephaly is connected to an OFC more than 3SD below the mean. Microcephaly is associated with a reduction in brain volume and frequently intellectual and/or motor inabilities. The pathogenesis of microcephaly is heterogeneous, extending from hereditary causes to environmental components that can have an effect on developmental process that impact brain size[1, 2].

Any condition that influences imperative processes of brain development, such as progenitor cell multiplication, cell differentiation, and cell death, can hence induce microcephaly. Irregularities leading tomicrocephaly may only influence cerebral development (non-syndromal microcephaly) or may be related with extracranial malformations and/or facial dysmorphism (syn-dromal microcephaly). Microcephaly may be apparent at birth (essential micro-cephaly) or postnatally (auxiliary microcephaly). The child with auxiliary microcephaly includes a typical OFC at birth and after that subsequently the relative OFC drops to avalue more than 2SD below the mean. These terms do not imply distinct aetiologies. Both primary and secondary microcephaly can be acquired or hereditary. The distinction of essential and auxiliary microcephaly empowers clinicians to rank the probability of a putative conclusion concurring to disease prevalence.

The phenotype of microcephaly is variable and the spectrum of related disorders is large, with more than 900 entries within the Online Mendelian Legacy in Man com-pendium for the clinical sign 'microcephaly' as of January 2014. The main point of study was to examine in a large cohort of patients with microcephaly (1) the recurrence of (putative) causes of microcephaly; (2) the recurrence of structural brain variations from the norm, mental inability, and associated clutters; (3) the demonstrative steps taken to define the fundamental malady; and (4) the number of cases in which the symptomatic approach was effective. We too propose uniform information documentation and a standardized initial diagnostic approach to children with microcephaly[3, 4].

MATERIAL AND METHODS

This is a quantitative cross sectional study arrangement based on 12 persons. On the basis of microcephaly, the subjects were separated into the two groups: Group I: Subjects with microcephaly (n=10), Group II: Normal kin as controls (n=2). Five families including add up to of 12 individuals was selected. Ten people with microcephaly (cases) and two normal kin (without microcephaly) were taken as controls. Cooperation of the subjects in this study was selected voluntarily and written informed consent was taken to take part in the study from each individual and from their guardians. The individuals and their guardians were educated about the potential benefits and risks of this study. An organized survey was filled by the primary examiner. Body weight (BW) and height of all members was recorded and Body mass index (BMI) was calculated as (BMI = BW (kg) / stature (m)2. Five ml blood of all members was drawn from the cubital vein through aseptic measures. The blood tests were centrifuged for 10 min and the serum sample was collected and put away at -80o C until utilized. All biochemical parameters were decided in duplicate utilizing standard procedures. Serum leptin, cortisol, development hormone (hGH) and thyroid stimulating hormone (TSH) concentrations were determined by chemical connected immunosorbent assay (ELISA) utilizing commercial kits. The importance of contrasts between two groups was analyzed by Free Tests Mann–Whitney U test. P-value <0.05 was considered factually significant. Information was entered and all calculations were carried out with the SPSS) version 23.

RESULTS

There was little difference within the mean age of microcephaly individuals as compared to healthy siblings. BMI of microcephaly

patients was 18.2 ±1.5 and of controls it was 25.5± 3.5, noteworthy affiliation was found between BMI and microcephaly. Mean serum leptin concentrations in microcephaly patients was 5.0 ±0.9 vs 22.5 ±11.5 compared with controls but the affiliation was non-significant. Though, mean cortisol levels in microcephaly patients were tall as compared with normal kin (30.5 ± 3.5 vs 24.5± 7.5) but the difference isn't noteworthy. No significant affiliation was found in serum development hormone and TSH levels in microcephaly patients as compared with controls. P-value was non-significant for leptin, cortisol, GH and TSH.

Table 1: Parameters of Age, BMI, Leptin, GH TSH and Cortisol between microcephaly and normal individuals.

No.	Parameter	Microcephaly patients (10)	Normal ones (n=2)	p-value
1.	Age	20.5 + 2.5	25.5 + 3.5	0.80
2.	BMI	18.2 + 1.5	25.3 + 3.0	0.04
3.	Leptin	5.0 + 0.9	22.5 + 11.5	0.10
4.	TSH	2.4 + 0.5	2.1 + 0.5	0.9
5.	GH	1.3 + 0.3	1.9 + 0.4	0.11
6.	Cortisol	30.5 + 3.5	24.5 + 7.5	0.85

P value ≤ 0.05 is significant

DISCUSSION

This study highlights a noteworthy affiliation between BMI and microcephaly patients. Our results are supported by another study on microcephaly patients which reported lower weight and height in addition to abnormal brain size. Peptides secreted by the fat tissue i.e., leptin and adiponectin, play a vital part in use of vitality and body weight control. Rett disorder, a neurological abnormality is characterized by microcephaly, development failure and behavioral disorders. So an affiliation can be proposed between leptin levels and microcephaly. There is no significant correlation is found in our study[5].

Another study on four microcephalic kin (3 guys and 1 female) appeared that they were brief heighted and had hypergonadotropic hypogonadism along with numerous congenital variations from the norm. This family moreover had 7 healthy kin. The affected kin moreover had impaired development and sexual developmental. Test results appeared ordinary development adrenocortical hormones and abnormal levels of sex hormones. Our study also appeared no critical affiliation between microcephaly and GH.A research on patients with low glucocorticoid levels uncovered neurological deficit in them. But in our study no significant affiliation was seen between cortisol and microcephaly[6].

A case report in 1991 included 2 cases of microcephaly, which appeared GH lack with abnormal arginine and insulin provocation tests[7]. An uncommon case was reported, who had a combination of short stature, impaired sexual improvement, microcephaly, hypothyroidism and altered pancreatic capacities. His chromosomal examination appeared 47 XXY13. These results might too demonstrate a few relationships of microcephaly and hypothyroidism. A research study on 3 female enduring from a disorder highlighted short stature, deafness, mental impediment, and tooth anomaly, microcephaly and thyroid dysfunction. But this study showed no such affiliation[8-10].

CONCLUSION

In light of this study it can be recommended that there's a significant affiliation of BMI and microcephaly but the affiliation of Leptin, Cortisol, GH and TSH with microcephaly seem not be found as proposed by non-significant results. This may moreover emphasize on heredity perspective of this condition. Limitations included less subjects hence study results cannot be generalized. Further studies ought to be done to decide the cause-effect relationship between endocrine profile and microcephaly.

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