

MICRO SYNDROME IN AN ADULT

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ARTICLE INFO

Article History:

Received 17th January, 2017

Received in revised form 8th February, 2017

Accepted 22nd March, 2017

Published online 28th April, 2017

Key words:

Micro syndrome, micro cornea, congenital cataract, micro genitalia

ABSTRACT

Any Patient who presents with congenital cataract in the younger age group needs a thorough clinical examination. Our patient had mental retardation with micro ophthalmia, micro cornea, and micro genitalia, operated for congenital cataract in the younger age. So a diagnosis of micro syndrome was made due to the predominant ocular manifestations.

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INTRODUCTION

Case report

A twenty year old female came to the medical outpatient department for the treatment of gastritis. She had past history of cataract, both eyes got operated for cataract at the age of two years. There was history of delayed motor and mental milestones since childhood. She attained menarche at the age of fourteen years, periods were irregular initially and with a present history of amenorrhoea for the past four years. On clinical examination She had high arched palate, mental retardation, stunted growth, short stature, under weight, micro ophthalmia, small cornea-micro cornea (9 mm in both meridians),

(figure.1) both eyes-aphakic (bilateral congenital cataract operated) with small head-microcephaly. The secondary sexual characters, and external genitals were normal. Investigations revealed Hb-9 gms, blood hemogram-microcytic hypochromic anemia. Thyroid function tests, renal function tests, blood sugar, urine examination, ECG, serum calcium-were all within normal limits. CT-brain-normal. No evidence of intracranial calcification or agenesis of corpus callosum. Buccal smear for Barr body-positive. Audiogram - normal study. Urine for galactose-negative. Ultrasonogram and CT scan of abdomen revealed small atrophied uterus and absent ovaries), (figure .2)



Figure 1 microphthalmia and micro cornea

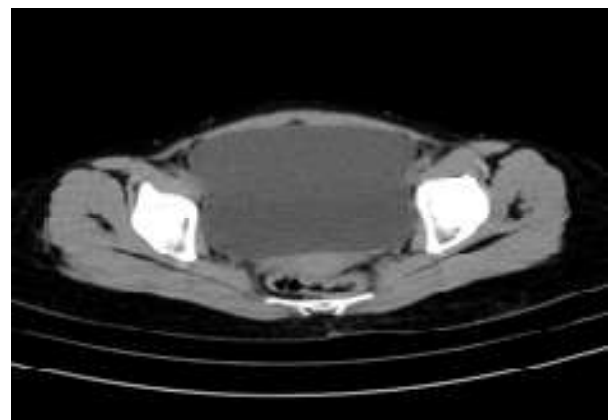


Figure 2 Atrophied uterus & absent ovaries

Since she had Mental retardation, congenital cataract, micro cornea, microgenitalia a diagnosis of micro syndrome was made.

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DISCUSSION

Micro syndrome was first described by Warburg (1). Micro syndrome is more commonly seen in children than in adults.(2). Micro syndrome is an autosomal recessive disorder characterized by ocular and neuro developmental defects and microgenitalia. (3) The pathology is due to abnormal neurotransmitter vesicular transport and exocytosis. In the literature few cases of micro syndrome were reported in children and younger age group rather than adults. (4,5)

Ocular findings are the most reliable diagnostic signs of micro syndrome. The clinical features are as follows -congenital cataract, microcornea, microphthalmia, microcephaly, mental retardation, atonic pupils, optic atrophy, and corpus callosum hypoplasia.(1). Congenital cataract is the most common symptom of the disease. Our patient had congenital cataract in childhood and it was operated in the younger age. Other clinical features include dysmorphic facies, high arched palate and ear abnormality which were present in our patient.

The causes of congenital cataract are viral infection-Rubella, sporadic, galactosemia, hypoparathyroidism, Down's syndrome, Trisomy syndromes, Cri du chat syndrome, Micro syndrome, toxins, drugs, irradiation neurofibromatosis, juvenile chronic arthritis and steroid therapy. The Differential Diagnosis for Micro syndrome are COFS syndrome-cerebro-oculo-facio-skeletal syndrome-severe brain atrophy and calcification can occur. (6, 7). NHS -Nance Horan Syndrome (XLD)-dental and facial anomalies are present, mental retardation can be seen in most of the cases. She was clinically diagnosed to have micro syndrome and was treated symptomatically and she is under regular follow up.

CONCLUSION

Our patient had clinical features suggestive of micro syndrome in the form of microcephaly, microphthalmia, microcornea, and microgenitalia. Ocular finding are the most reliable diagnostic signs of micro syndrome. So micro syndrome should be considered in any patient with congenital cataract and microphthalmia and micro cornea with mental retardation.

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How to cite this article:

Kannan R *et al* (2017) ' Micro Syndrome In An Adult ', *International Journal of Current Advanced Research*, 06(04), pp. 3102-3103. DOI: <http://dx.doi.org/10.24327/ijcar.2017.3103.0193>
