OCULAR FINDINGS IN COMBINED METHYLMALONIC ACIDEMIA AND HOMOCYSTINURIA

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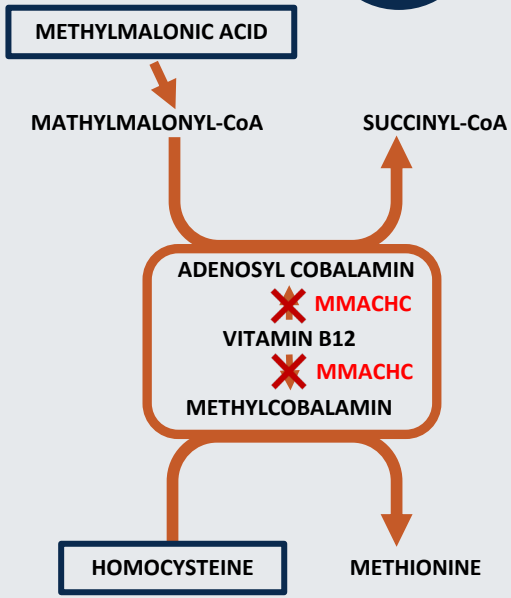
RETINA 2023
47th BRAVS MEETING
APRIL 28th to MAY 1st

PURPOSE

METHYLMALONIC ACIDURIA AND HOMOCYSTINURIA, CBLC TYPE

COMBINED METHYLMALONIC ACIDEMIA AND HOMOCYSTIURIA COBALAMIN C (cblC) TYPE, IS A CONDITION THAT OCCURS SECONDARY TO **IMPAIRED CONVERSION OF DIETARY COBALAMIN (VITAMIN B12) TO ITS METABOLICALLY ACTIVE FORMS** DUE TO MUTATIONS IN THE ***MMACHC*** **GENE**. IT CAN MANIFEST CLINICALLY AS **FAILURE TO THRIVE**, **MEGALOBLASTIC ANEMIA**, **NEUROLOGIC DYSFUNCTION** AND **OPHTHALMIC OMPLICATIONS**.

WE REPORT THE CASE OF A PEDIATRIC PATIENT WITH METHYLMALONIC ACIDEMIA AND HOMOCYSTIURIA cblC TYPE DIAGNOSED ON NEWBORN METABOLIC DISEASE SCREENING, AND DESCRIBE THE ASSOCIATED OCULAR FINDINGS



METHODS

14-DAY-OLD FEMALE NEWBORN

REFERRED BY THE NEONATAL SCREENING UNIT WITH SUSPECTED METHYLMALONIC ACIDEMIA WITH HOMOCYSTINURIA. SHE PRESENTED WITH FAILURE TO THRIVE, DEHYDRATION, HYPOTONIA AND LABORATORY FINDINGS OF MARKEDLY RAISED SERUM HOMOCYSTEINE AND URINARY METHYLMALONIC ACID LEVELS. **PROMPT GENETIC TESTING CONFIRMED CBLC DISEASE**. THE PATIENT INITIATED TREATMENT WITH HYDROXYCOBALAMIN, COMBINED WITH BETADINE AND FOLIC ACID AND **MAINTAINED REGULAR OPHTHALMIC FOLLOW-UP.**

RESULTS

SEQUENTIAL OPHTHALMIC EXAMINATIONS WERE **NORMAL UNTIL THE AGE OF 2 YEARS** WHEN A **PENDULAR, HORIZONTAL NYSTAGMUS** WAS FIRST NOTED, FOLLOWED BY **POOR VISUAL FIXATION AND COMPROMISED PURSUIT OCULAR MOVEMENTS**

ANTERIOR SEGMENT OBSERVATION OU WAS CONSISTENTLY UNREMARKABLE

RESULTS

RETINA 2023
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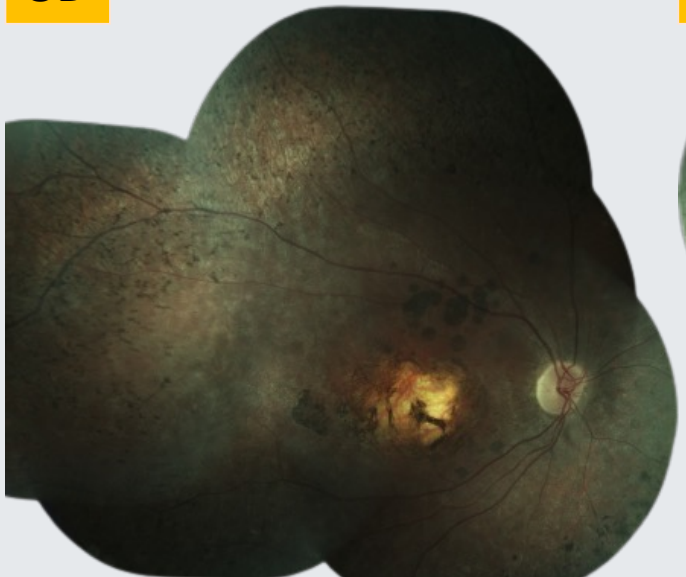
BY AGE 9 BCVA WAS 20/50 OU AND OPHTHALMIC EXAMINATION REVEALED:

1 DILATED FUNDUS EXAMINATION

- AREA OF MACULAR CHORIORETINAL ATROPHY WITH RETINAL PIGMENTED EPITHELIUM INVOLVEMENT INVOLVING THE FOVEA
- SURROUNDING HYPERPIGMENTED LESIONS
- DISCRETE OPTIC NERVE PALLOR AND ARTERIOLAR NARROWING

IMAGE 1. FUNDUS RETINOGRAPHY AT 9-YEARS-OLD

OD



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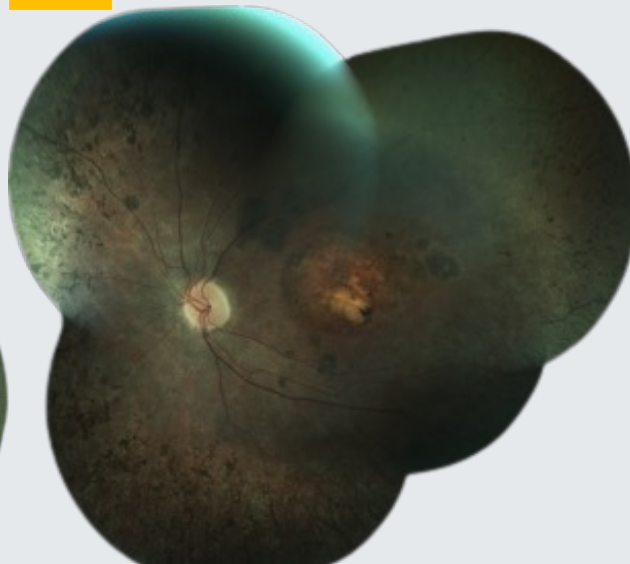


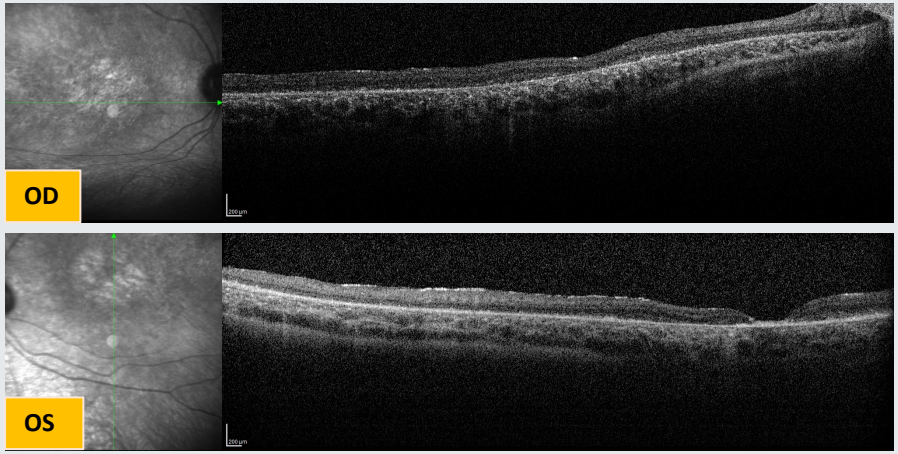
RESULTS

BY AGE 9 BCVA WAS 20/50 OU AND OPHTHALMIC EXAMINATION REVEALED:

2 SD-OCT

COMPROMISED BY LACK OF PATIENT COOPERATION AND THE PRESENCE OF NYSTAGMUS, BUT A **LOSS OF THE NORMAL RETINAL CYTOARCHITECTURE WITH AREAS OF ATROPHY AND CHOROIDAL HYPER-TRANSMISSIBILITY** ARE APPARENT.

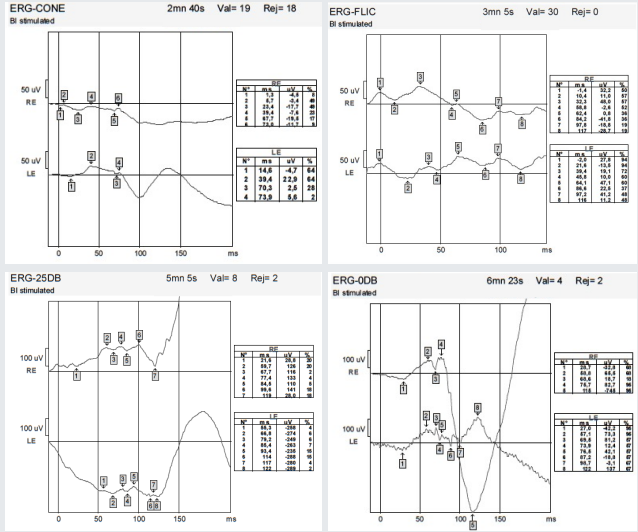
IMAGE 2. SD-OCT AT 9-YEARS-OLD



3 ELECTRORETINOGRAPHY (ERG)

SCOTOPIC AND PHOTOPIC RESPONSES ARE REDUCED AND DELAYED

IMAGE 3. ERG AT 9-YEARS-OLD



CONCLUSION

THE PRESENT CASE DESCRIBES THE EVOLUTION OF THE OCULAR MANIFESTATIONS IN A CHILD WITH A CONFIRMED DIAGNOSIS OF TYPE CBLC METHYLMALONIC ACIDEMIA WITH HOMOCYSTINURIA

DESPITE SOME PHENOTYPIC VARIABILITY, OCULAR FEATURES WITH PROGRESSIVE IMPAIRMENT OF VISUAL ACUITY ARE EXPECTED DURING THE FIRST DECADE OF LIFE

THIS DISEASE SHOULD BE KEPT IN MIND IN THE DIFFERENTIAL DIAGNOSIS OF INFANTILE MACULOPATHY, ESPECIALLY WHEN THE NEWBORN METABOLIC SCREENING IS NOT AVAILABLE OR PERFORMED

ALTHOUGH THE METABOLIC SCREENING ALLOWS FOR A PROMPT TREATMENT THAT CAN SIGNIFICANTLY INCREASE THE PATIENTS' LIFESPAN, IT REMAINS TO BE EXPLAINED WHY THE OCULAR MANIFESTATIONS ARE UNREMITTING