ADA2 DEFICIENCY PRESENTING AS INFANTILE POLYARTERITIS NODOSA

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Background: Polyarteritis nodosa (PAN) is a rare form of primary systemic vasculitis with heterogeneous presentations, treatments and disease course. There have been improvements in understanding of certain causes of PAN such as the association of the monogenic disorder Deficiency of Adenosine Deaminase 2 (DADA2) with a PAN phenotype. This described autoinflammatory disease is caused by loss-of-function homozygous or compound heterozygous mutations in CECR1 and can be characterised by an early onset vasculopathy with clinical and histopathological features of PAN associated with haemorrhagic and ischaemic strokes.

Objectives: We report a case of a 14 year-old boy, with a prenatal diagnosis of Klinefelter syndrome, whose parents are first cousins. We report a case of mutant adenosine deaminase 2 in a polyarteritis nodosa patient.

Methods: Review of patient’s clinical records and scientific literature.

Results: During his first two years of age, he suffered two small strokes, which confirmed our suspicion. He completed induction treatment with IV cyclophosphamide (6 months), steroids, methotrexate. Biological therapy was administered to 2 patients: 1 received belimumab and then rituximab, while the other patient received rituximab. All patients needed plasmapheresis and immunosuppressive therapy with cyclophosphamide. Biological therapy was administered to 2 patients: 1 received belimumab and then rituximab, while the other patient received rituximab.

Conclusion: The age of onset and clinical manifestations of DADA2 vary widely and are not clearly related to the genotype. Although this variant in CECR1 was of uncertain pathological significance, the absence of ADA2 enzymatic activity confirms the diagnosis. Timely diagnosis and treatment with anti-TNF drugs are thought to be crucial to prevent severe complications. Also, these patients should not be treated with ASA or anticoagulants which are frequently used in PAN patients. Low IgM levels can be a clue to raise the suspicion. Lack of long term outcome data in children with DADA2 makes the prognosis uncertain and underlines the need to maintain a lifelong follow-up.

REFERENCES

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