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Expanding the Clinical Spectrum of *Deficiency of Adenosine Deaminase 2 (DADA2)* : A Case of Conus Cauda Syndrome

Shivam Mirg [1], Ayush Agarwal[1], Awadh K Pandit [1], Divya MR [1], Divyani Garg [1], Ajay Garg [2], Achal Kr Srivastava [1]

[1]-Department of Neurology
[2]-Department of Neuro-Radiology
All India Institute of Medical Sciences, New Delhi

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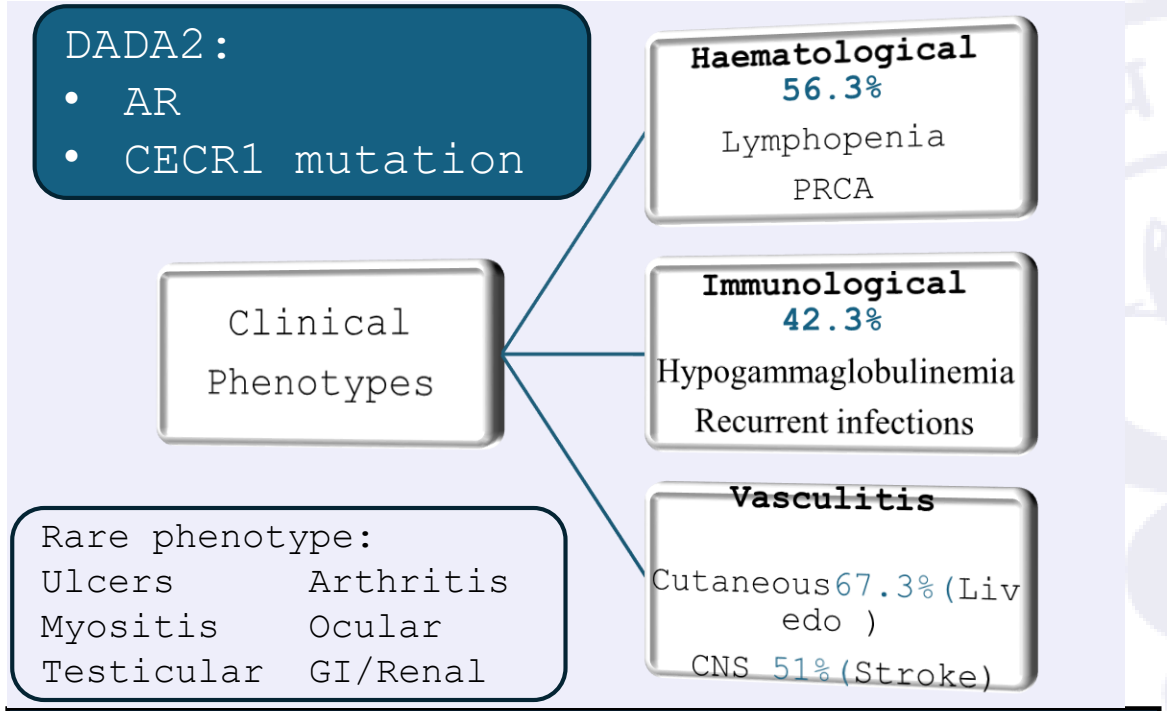

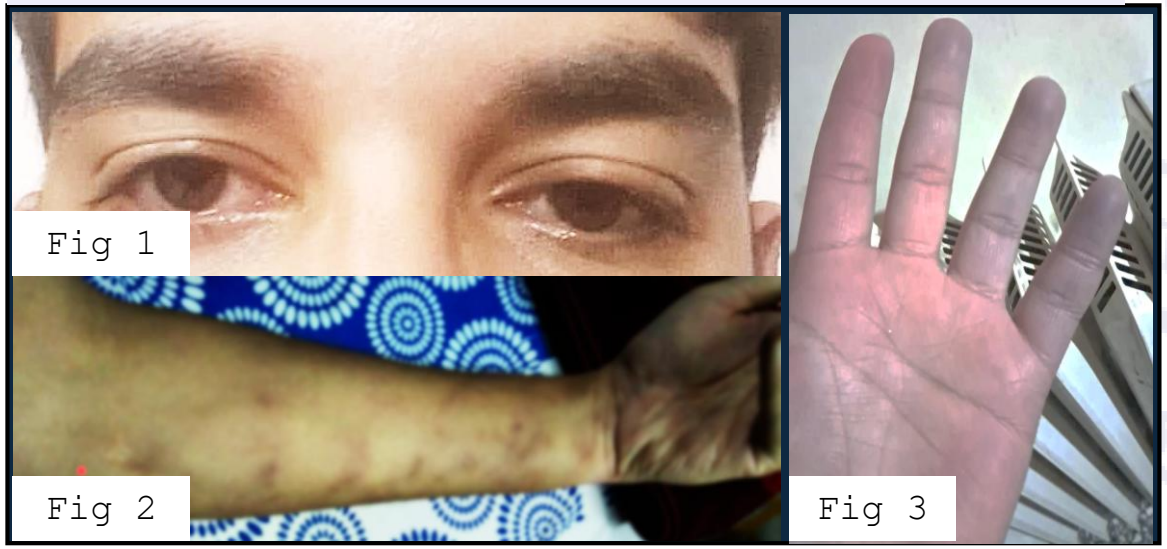
Fig 1

Fig 2

Fig 3

METHODS & MATERIALS

- 27-year-old Male
- **Two-week history** of acute onset , rapidly progressive, asymmetric, left > right paraparesis, along with bowel and bladder incontinence.
- **Examination** revealed proximal > distal weakness in the left leg (hip 2/5, ankle 3/5), brisk knee reflexes, absent ankle reflex, extensor plantar on the left, and saddle anaesthesia.
- **Past history** included young-onset hypertension, Headache with recurrent 3rd nerve palsy (self limiting; Fig1) , Raynaud phenomenon (Fig2) , and livedo racemosa (Fig3).
- **Family history** of stroke in 5th decade in paternal grandparents and paternal aunt & uncle

RESULT

- **Syndromic diagnosis**: Myeloradiculopathy
- **Differentials**: Vasculitides (e.g., PAN, lupus, Sjögren's, ANCA-associated), infections (CMV, HTLV, HSV, syphilis), and malignancy (lymphoma/metastasis).
- **MRI** revealed asymmetric thickening/enhancement of left lumbosacral nerve roots (Fig 4).
- **CSF** was acellular with normal protein/glucose and no malignant cells.
- **PET scan** showed no abnormal uptake.
- **Autoimmune and vasculitis profiles** were negative.
- **Whole exome sequencing** revealed a **homozygous missense mutation** in *ADA2* (c.139G>A; p.Gly47Arg), confirming DADA2.
- **Treatment:** Pulse Steroids with intravenous immunoglobulin & cyclophosphamide maintenance.
- **Follow-up:** Near complete recovery at 3-month

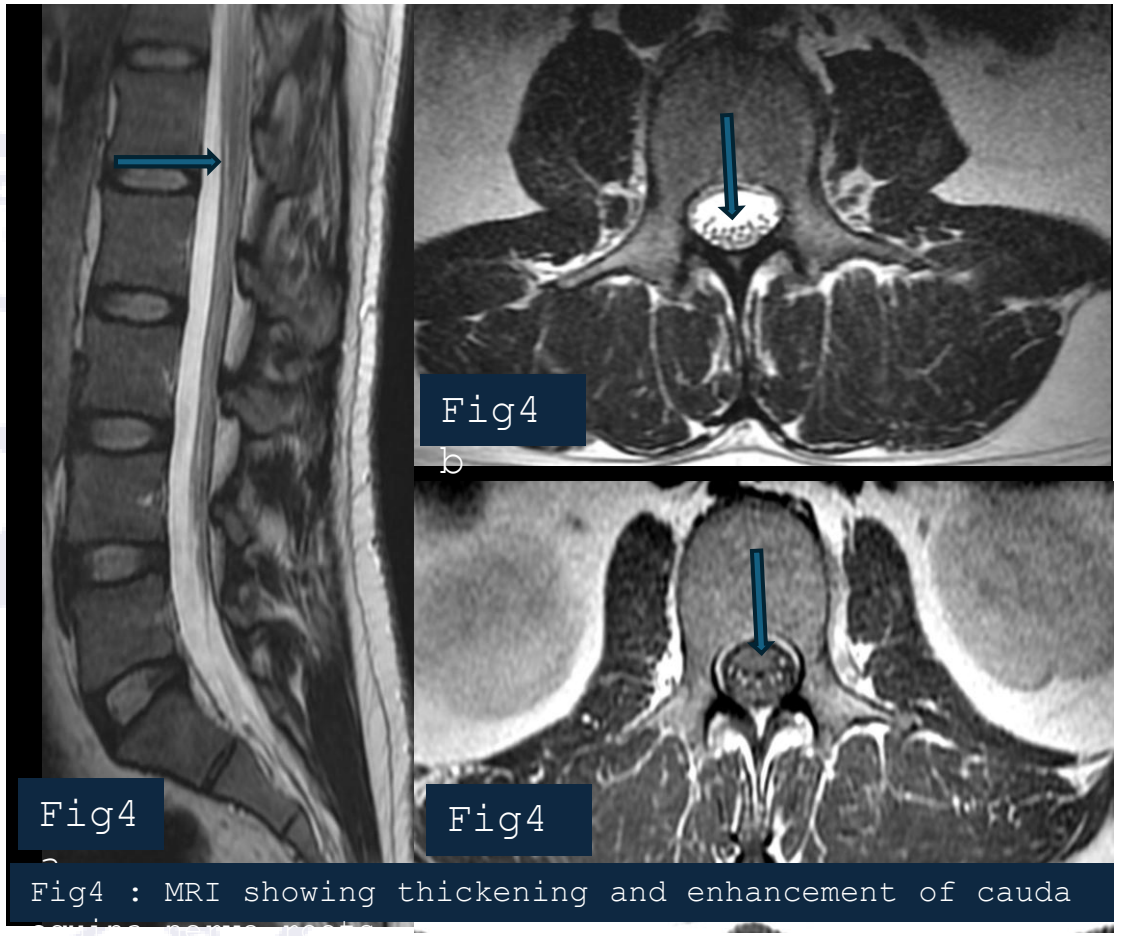

Fig4 : MRI showing thickening and enhancement of cauda

CONCLUSION

- This case expands the known phenotypic spectrum of DADA2 to include conus cauda syndrome, recurrent self limiting 3rd cranial nerve palsy, migraine like headaches —features rarely reported.
- Genetic testing for DADA2 should be considered in all suspected cases of early-onset or familial cases of PAN.
- Phenotypic heterogeneity in DADA2 is well recognized.
- Increased awareness among specialists is essential for timely diagnosis and management.

References

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THANK YOU

For any further queries mail us at
shivammirg@aiims.edu

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