Amyloidosis in Charcot Marie Tooth Disease – A rare complication

Antony Judy M[1], Nidhi Rawat[2]

Department of Physical Medicine and Rehabilitation, St John's Medical College Hospital, Bangalore, India.

INTRODUCTION

- ✓ **Charcot Marie Tooth disease (CMTD) is the most common inherited peripheral neuropathy 1**
- ✓ Amyloidosis is a rare complication of CMTD
- ✓ Kidney is the most common affected organ Amyloidosis.[2]

CASE REPORT

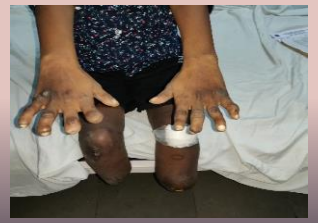
✓ A 17 year old female with CMTD - Type II, auto-amputation of digits, and trans-tibial amputation (figure) who is independent in all ADLs and ambulating with bilateral trans-tibial prosthesis.

✓ **Chief Complaints:**

Intermittent fever, difficulty in breathing since five days, progressive swelling of abdomen, frothy urine, and wound over distal end of left residual limb since five years.

✓ **Examination:**

- Tachycardia.
- Bilateral pitting oedema up to mid thigh
- Ascites.
- Ulcer over the distal end of left residual limb.



INVESTIGATIONS

- ✓ **Blood:** TC – **16.09**, Creatinine – **5.33**, TP/Albumin – **3.8/1.1** , TSH – **20.49**, C3/C4/AA – **Negative**.
- ✓ **USG ABD:** Liver - 11.5cm, ascites, minimal pleural effusion and **chronic cystitis**.
- ✓ Cystitis was treated with antibiotics. However, as systemic symptoms did not resolve the case was discussed with Nephrology.
- ✓ **Renal biopsy: PAS stain positive, Congo stain positive and apple green bifringence suggestive of renal amyloidosis**.

TREATMENT

- ✓ Two cycles of haemodialysis were given, following which her symptoms resolved and her renal function normalised

DISCUSSION

- ✓ Amyloidosis is a group of disorders in which soluble proteins aggregate and deposit extracellularly in tissues causing organ dysfunction.[2]
- ✓ The **risk factors** for amyloidosis are age related, familial, and **chronic infection/ inflammation**.[2]
- ✓ Renal Amyloidosis if left untreated can lead to end stage renal disease (ESRD).[2]
- ✓ Yoshitaka Kikukawa et al reported a similar case in an elderly lady with CMTD who responded to treatment with chemotherapeutic agents. [3]
- ✓ **In our case, chronicity of ulcer and cystitis are the probable risk factors leading to Amyloidosis and associated clinical features**

CONCLUSION

- ✓ **This rare case highlights the presence of Amyloidosis in a patient with CMDT due to chronic infection/inflammation**.
- ✓ CMDT patients with chronic wounds and repeated infections should be educated regarding the possibility of developing Amyloidosis.
- ✓ Amyloidosis is to considered in CMDT patients with chronic infection/inflammation unresponsive to routine infection treatment protocols

REFERENCES

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