# GENE THERAPY: A GLIMMER IN THE GLOOM OF METACHROMATIC LEUKODYSTROPHY (MLD)

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## BACKGROUND

- MLD is a rare but severe & progressive AR demyelinating condition of the CNS.
- Lysosomal enzyme - arylsulfatase A (*ARSA*) – deficiency ⟶ build-up of sulphatides in the nervous system ⟶ neurocognitive deterioration.
- At present, there is no cure for MLD albeit various therapeutic options are being explored. One of such therapeutic options is gene therapy.

## CASE REPORT

- Herein we report the case of an 18 month old girl of healthy non consanguineous parents.
- She presented with rapid neurocognitive deterioration, MRI findings & genetic testing consistent with late infantile MLD.
- With symptomatic MLD & no prospect of gene therapy, she died at the age of 5 years.
- However, the onset of MLD in our patient prompted the investigations & diagnoses of presymptomatic MLD in her 3 month old monozygotic twin brothers.
- The twins received autologous bone marrow transplant with cells transduced with ARSA encoding lentiviral vector to good effects.
- Post-transplant course has been uneventful save for an episode of hepato veno-occlusive disease in one of the twins.
- Currently at the age of 35 months, the twins are still asymptomatic of MLD.

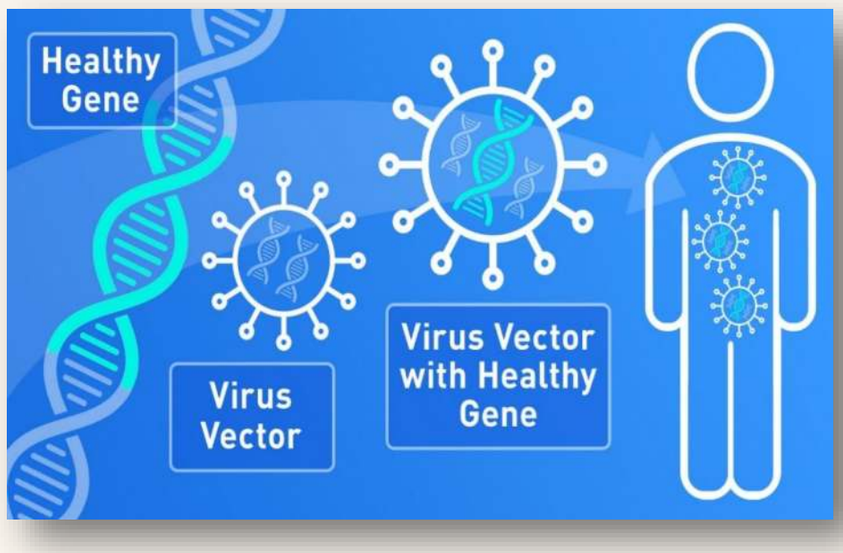


*Fig 1: Gene therapy infographic*

## CONCLUSION

- Our case report attests to the potential of gene therapy in the treatment MLD.
- However, further research is needed to study risk and long term outcomes in these patients.

## References

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