The reported patient presented with atypical features of CAPS with elevated acute phase response and mutations in NLRP3 gene. Cryopyrin-associated periodic syndrome (CAPS) is a rare inflammatory disorder with three phenotypes: Familial cold autoinflammatory syndrome (FCAS), Muckle-Wells syndrome (MWS) and neonatal-onset multisystem inflammatory disease (NOMID). Among them, NOMID type is the severe form presenting with central nervous system involvement and commonly arises from new mutations (2). The clinical manifestations in our reported patient showed predominantly CNS involvement with headache, papilloedema, raised intracranial pressure and sensorineural hearing loss, in keeping with atypical NOMID type and a coexisting new mutation.

Learning Points/Conclusion: We report the first case of a child with Phe566Tyr mutation, phenotypically presenting as atypical NOMID type of CAPS with papilloedema, hearing loss and elevated acute phase response. Also in our patient, as previously reported (3), showed marked improvement with Anakinra treatment.

References
2) Yu J.R., Leslie K.S. Cryopyrin-Associated Periodic Syndrome: An Update on Diagnosis and Treatment Response. Curr Allergy Asthma Rep (2011);11:12