literature. Ann Indian Acad Neurol. 2019;22:361-3.
2. Campbell H, Andrews N, Brown KE, Miller E. Review of the effect of measles vaccination on the epidemiology of SSPE. Int J Epidemiol. 2007;36:133448.
3. Saurabh K, Gupta R, Khare S, Sharma S. Atypical subacute sclerosing panencephalitis with short onset latency. Indian Pediatr. 2013;50:244-5.
4. Anlar B. Subacute sclerosing panencephalitis and chronic viral encephalitis. Handb Clin Neurol. 2013;112:1183-9.
5. Gutierrez J, Issacson RS, Koppel BS. Subacute sclerosing panencephalitis: An update. Dev Med Child Neurol. 2010;52:901-7.

Allgrove Syndrome and a Novel Mutation of *AAAS* Gene in a Boy

A 2.5-year-old boy presented with progressive darkening of lips for the past four months, and absence of tears noticed since early infancy. There was no history of dysphagia or neurological problems. Born to non-consanguineous parents, his two elder sisters had died at age 5 and 6 years after similar manifestations. On examination, he was underweight (9.2 kg, –3.07SDS), stunted (82.4 cm, –2.68SDS) and had small head (45.5 cm, –2.42SDS). Oral mucosa was deeply pigmented. The systemic examination was unremarkable. Laboratory investigations showed normal serum sodium and potassium but low morning serum cortisol (89.2 nmol/L) and elevated adrenocorticotropin levels (1470 pg/mL). Reduced tear production was confirmed by Schirmer test. Barium swallow and endoscopic evaluation showed no achalasia. Clinical exome sequencing showed a previously undescribed homozygous frameshift deletion c.762delC (p.Ser255Valfs* 36) at Exon 8 of the *AAAS* gene, further confirmed by Sanger sequencing. The mutation is considered pathogenic by prediction tools such as SIFT, Mutation Taster and Phenolyzer. The child was initiated on lubricant eye drops and hydrocortisone and showed improved growth over one year (weight 10.8 kg, –2.12 SDS and height 89.4 cm, –2.54 SDS).

Allgrove syndrome (or Triple A syndrome) is an autosomal recessively inherited disorder caused by mutations in the *AAAS* gene that encodes for a protein ALADIN involved in the movement of molecules into and out of the nucleus, probably affecting DNA repair mechanisms leading to cell death [1]. The syndrome may present with any one of the four cardinal features that include achalasia, Addison disease, alacrima and progressive neurological dysfunction, and the symptoms may evolve over a period of time [1]. Alacrima is often the earliest and most consistent finding. Presence of one more symptom warrants molecular analysis of the *AAAS* gene [2]. Although a lower prevalence of neurological dysfunction has been noted in Indian patients, this feature is known to manifest later [1]. The poor head growth in our patient probably indicates the beginning of neurological dysfunction. These patients are also prone to dermatological abnormalities such as hyperkeratosis. The *AAAS* gene mutations are known to affect siblings and may explain sibling deaths with adrenal failure [1,3]. A high index of clinical suspicion is required due to the rarity and presentation as incomplete triad of symptoms [4,5]. Molecular analysis of the *AAAS* gene helps in confirming diagnosis and prognostication.

Contributors: SG: data collection and literature search; DD: drafting of manuscript. The final version of the manuscript was approved by all authors.
Funding: None; *Competing interests*: None stated.

SANIYA GUPTA AND DEVI DAYAL*
Department of Pediatrics,
Advanced Pediatrics Center,
Postgraduate Institute of Medical Education and Research,
Chandigarh, India.
**drdevidayal@gmail.com*

REFERENCES

1. Patt H, Koehler K, Lodha S, Jadhav S, Yerawar C, Huebner A, *et al*. Phenotype-genotype spectrum of AAA syndrome from Western India and systematic review of literature. Endocr Connect. 2017;6:901-13.
2. Kurnaz E, Duminuco P, Aycan Z, Savaº-Erdeve S, Muratoolu Þahin N, Keskin M, *et al*. Clinical and genetic characterisation of a series of patients with triple A syndrome. Eur J Pediatr. 2018;177:363-9.
3. Bouliari A, Lu X, Persky RW, Stratakis CA. Triple A syndrome: Two siblings with a novel mutation in the AAAS gene. Hormones (Athens). 2019;18:109-12.
4. Shah SWH, Butt AK, Malik K, Alam A, Shahzad A, Khan AA. AAA syndrome: Case report of a rare disease. Pak J Med Sci. 2017;33:1512-6.
5. Reimann J, Kohlschmidt N, Tolksdorf K, Weis J, Kuchelmeister K, Roos A. Muscle pathology as a diagnostic clue to allgrove syndrome. J Neuropathol Exp Neurol. 2017;76:337-41.