

Riana RASAMISON, Jean-Meidi ALILI, Marie-Pierre BERLEUR, Marie-Caroline HUSSON

Regulatory, Pharmaceutical and Medical Department, Pharmaceutical Establishment of Paris Hospitals Group, AP-HP, Paris, France
 riana.rasamison@aphp.fr, marie-caroline.husson@aphp.fr

Background and Objective

Amino acids Isoleucine (ILE) and Valine (VAL) are used to treat Maple Syrup Urine Disease (MSUD). MSUD is a rare disease due to an enzyme deficiency which results in elevations of the branched-chain amino acids (ILE, VAL and Isoleucine) in plasma. Symptoms can lead to irreversible neurological complications, metabolic decompensation or death if untreated.

Aim of this work

→ To study the current therapeutic uses of two hospital preparations of ILE 50 mg capsules and VAL 50 mg capsules in French hospitals

Setting and Method

Main outcome measures

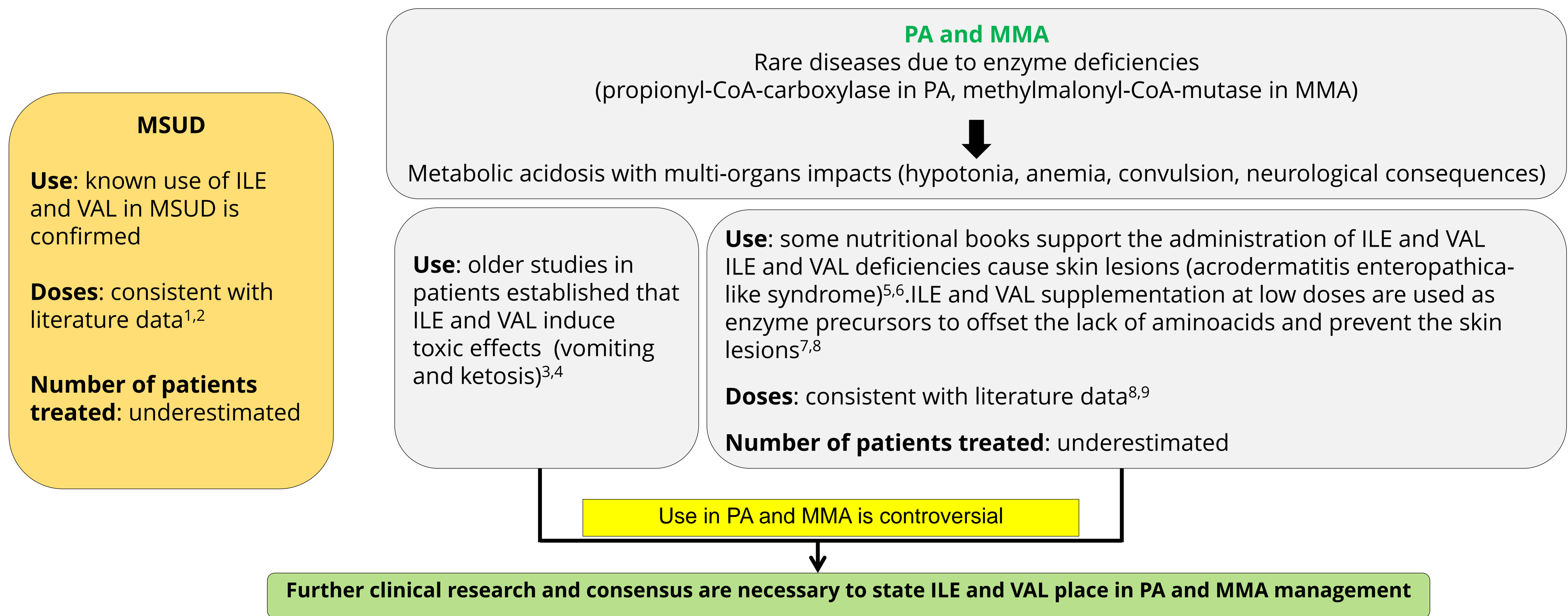


Results

Responses received from eight hospitals (n=8/17) including 4 reference centers for metabolic diseases (8 pharmacists and 4 clinicians)

ISOLEUCINE and VALINE	2 uses	Doses	Number of patients treated in 2016	Alternative treatments
KNOWN USE	MSUD	5 – 20 mg/kg/d (newborn) 600 - 1200 mg/d (adults)	65	Hospital preparation of capsules (ILE and VAL)
NEW USE IDENTIFIED	Propionic and MethylMalonic Acidemias (PA and MMA)	150 – 400 mg/d (children)	3	

Discussion



Conclusion

- ILE combined with VAL are indispensable to treat MSUD.
 - Further clinical research is required to assess if use of ILE combined with VAL in propionic and methylmalonic acidemias is indispensable.
 - Doses: they vary with the therapeutic uses and the severity of the symptoms. They are consistent with literature data.
- This survey has now to be completed with the total number of treated patients in France to identify a target population.