

Amino acid analysis outside the U.K.

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AAA working party

The logo for MetBioNet, featuring the text "MetBioNet" in a blue, serif font. The "Met" is on the top line, "Bio" is in the middle, and "Net" is on the bottom line. The letters are slightly overlapping and have a subtle shadow effect.

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How are patients investigated?

<u>LAB</u>	<u>Urine (U)</u>	<u>Plasma (P)</u>	<u>Paired U+P</u>
A	Qual HVE	No cold P req	
B	Quant if better marker than P	Majority P only	
C	Quant limited no.	Pref pre-lunch	U orgs
D	TMS – 2000 p.a.		
E	Qual TLC		
F	Quant limited no.	Majority P only	

Plasma aa's-what patient groups?

<u>LAB</u>	<u>Diagnosis</u>	<u>Monitoring</u>	<u>Nutrition</u>
A	Follow up abn urine	Yes	No
B	Yes	Yes	No
C	Yes	Yes	Eval new baby feeds
D	Yes	Yes	No
E	Yes	Yes	No
F	Yes	Yes	Yes

Reports

<u>LAB</u>	<u>Report</u>
A	Partial profiles
B	Levels & interpretive comment & conclusions FI will contact & discuss, not on report
C	Own Ref values, interpretation & advice PKU – partial profile, occasional full profile
D	Used to report all. Now qual report if all normal. Levels of significant/relevant aa's *

Reports - continued

<u>LAB</u>	<u>Report</u>
E	Interact with clinicians & offer appropriate additional tests. Rest of country uses MSMS - no clinical interaction
F	Interpretive comment even to specialist physician.* Compare with previous results

Technology

A	GCMS backup for IEC MSUD profiles
B	HPLC & Ninhydrin. MSMS still problematic.
C	TMS not ripe yet. Can't beat IEC.
D	No replacement for IEC yet. LC/MSMS close but needs fine tuning.
E	Love to know comparison between IEC & TMS!
F	Like all on TMS. Would lose extraneous info from aa profile & BUN. Working on home specimen BS paper collection

Other comments

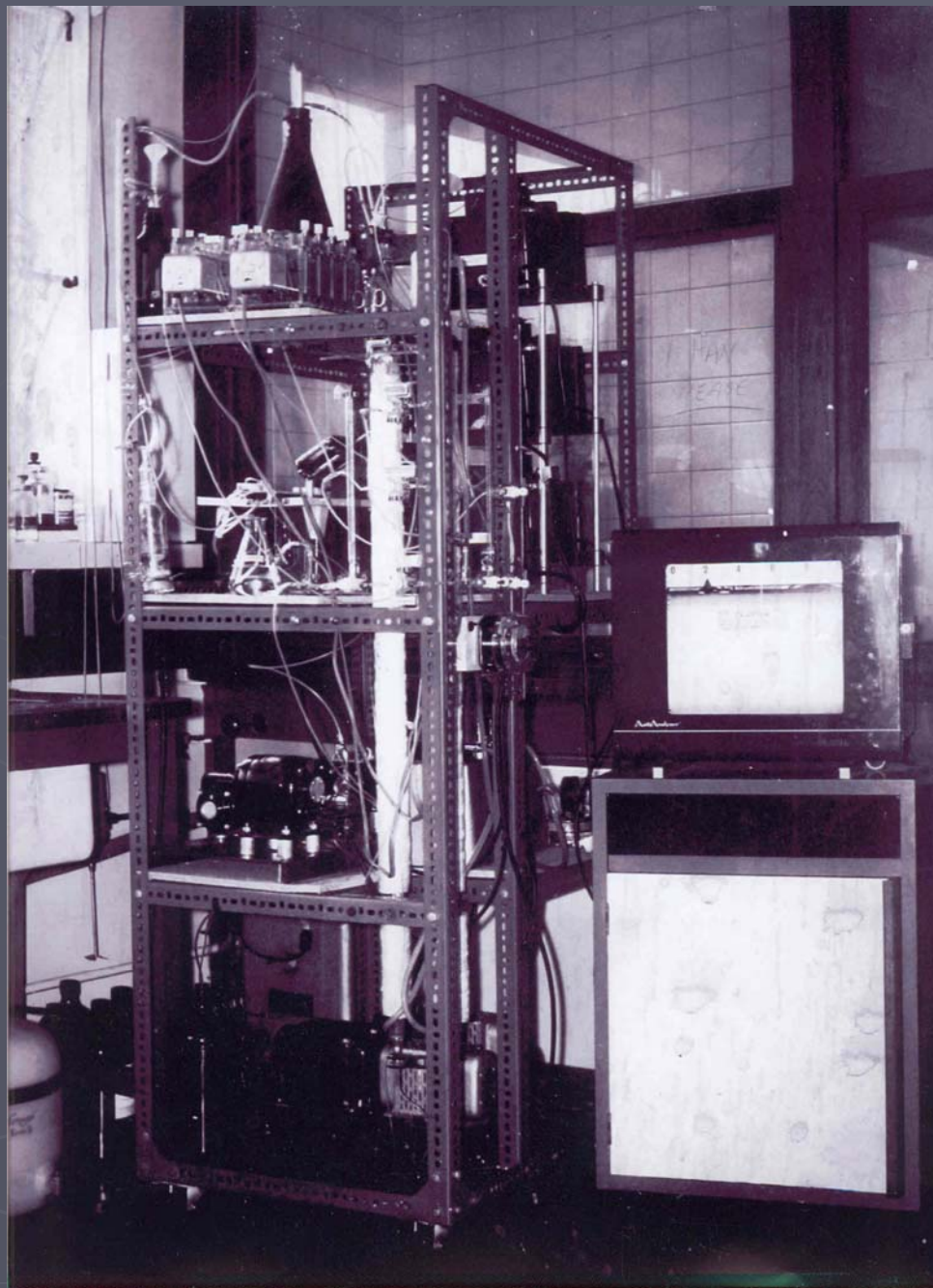
A	HVE old method but effective
B	Good recommendations from ERNDIM & ACMG
C	Bloodspot precision not good enough for unstable patients
D	Patients tested cheaper & more efficiently with urine TMS. Many IEC panels are overkill!
E	Quoted £100K for BIOCHROM!
F	CSF quant should include Ser, Gly, Gln & Thr minimum in early onset seizures & or microceph

Other comments - continued

4 USA labs; calibration & control expectations from lab inspectors:

- More than 1 level control daily
- Calib, Hi & Lo QC each run (24 hrs)
- Carryover issue
- Try to fit IEC quirks into general chemistry testing
- Checklists “force fit” into BCG practices

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