Expanded Newborn Screening positive for Multiple Acyl-CoA Dehydrogenase Deficiency?

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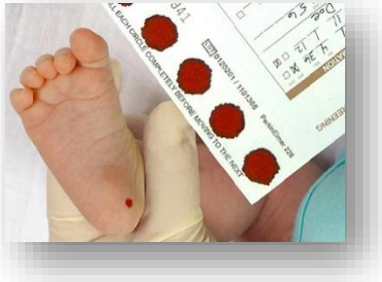
Newborn Screening results

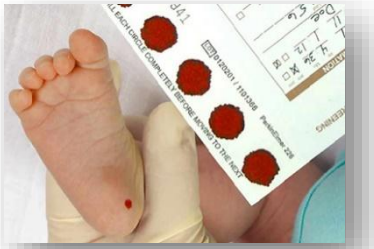


	Ptz 1		Ptz 2	
	SNE	recall	SNE	recall
C4 (<0.87)	0,96	1,04	1,29	1,78
C6 (<0.21)				0,27
C8 (<0.22)				0,56
C10 (<0.29)	0,31	0,08		0,55
C12:1 (<0.29)	0,43	0,05		0,75
C12 (<0.50)	0,62	0,15		0,87
C14:1 (< 0.4)	0,73	0,12		1,16
C14 (<0.6)	0,79	0,1		0,63
C16:1 (<0.6)				0,4
C16OH (<0.1)				0,09
Ethylmalonic acid (< 2,34)	14,9	100	11,1	170
Organic acid urine		Succinic acid + AGs*		glutaric acid + AGs*

MADD?

*AGs: Acylglycine (isobutylglycine; isovalerylglycine; hexanoylglycine

Newborn Screening results



	Patient 1			Patient 2		
	NBS	**recall**	**B2 100 mg/d**	**NBS**	**recall**	**B2 100 mg/d**
C4 (<0.87)	0,96	1,04	0,19	1,29	1,78	0,32
C6 (<0.21)					0,27	0,12
C8 (<0.22)					0,56	0,07
C10 (<0.29)	0,31	0,08	0,04		0,55	0,04
C12:1 (<0.29)	0,43	0,05	0,06		0,75	0,06
C12 (<0.50)	0,62	0,15	0,11		0,87	0,06
C14:1 (< 0.4)	0,73	0,12	0,1		1,16	0,08
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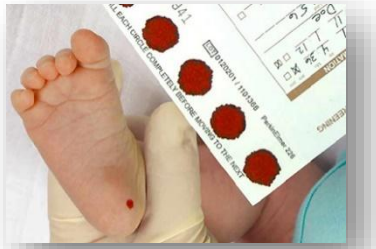
C16:1 (<0.6)					0,4	0,08
C16OH (<0.1)					0,09	0,06
EMA (< 2,34)	14,9	100	nd	11,1	170	nd
Organic acids U		succinic acid + AGs	Normal		glutaric acid+AGs	Normal

Molecular testing for MADD-related genes *ETFA, ETFB, ETFDH, FLAD1, SLC25A32, SLC52A1, SLC52A2, SLC52A3, ETHE1*: negative

Newborn Screening results MADD?



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EMA (< 2,34)	14,9	100	nd	11,1	170	nd
Organic acids U		succinic acid + AGs	Normal		glutaric acid+AGs	Normal
B2 (137-270 µ/L)		97	192		na	191
	Mother			**Mother**		
B2	121			113		

Maternal nutritional habits: vegan (patient 1), dairy-product restricted diet (patient 2)
Plasma riboflavin levels was decreased in both mothers: **MATERNAL B2 DEFICIENCY**

Newborn Screening results MADD?

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CASE REPORT

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Abnormal VLCADD newborn screening resembling MADD in four neonates with decreased riboflavin levels and VLCAD activity

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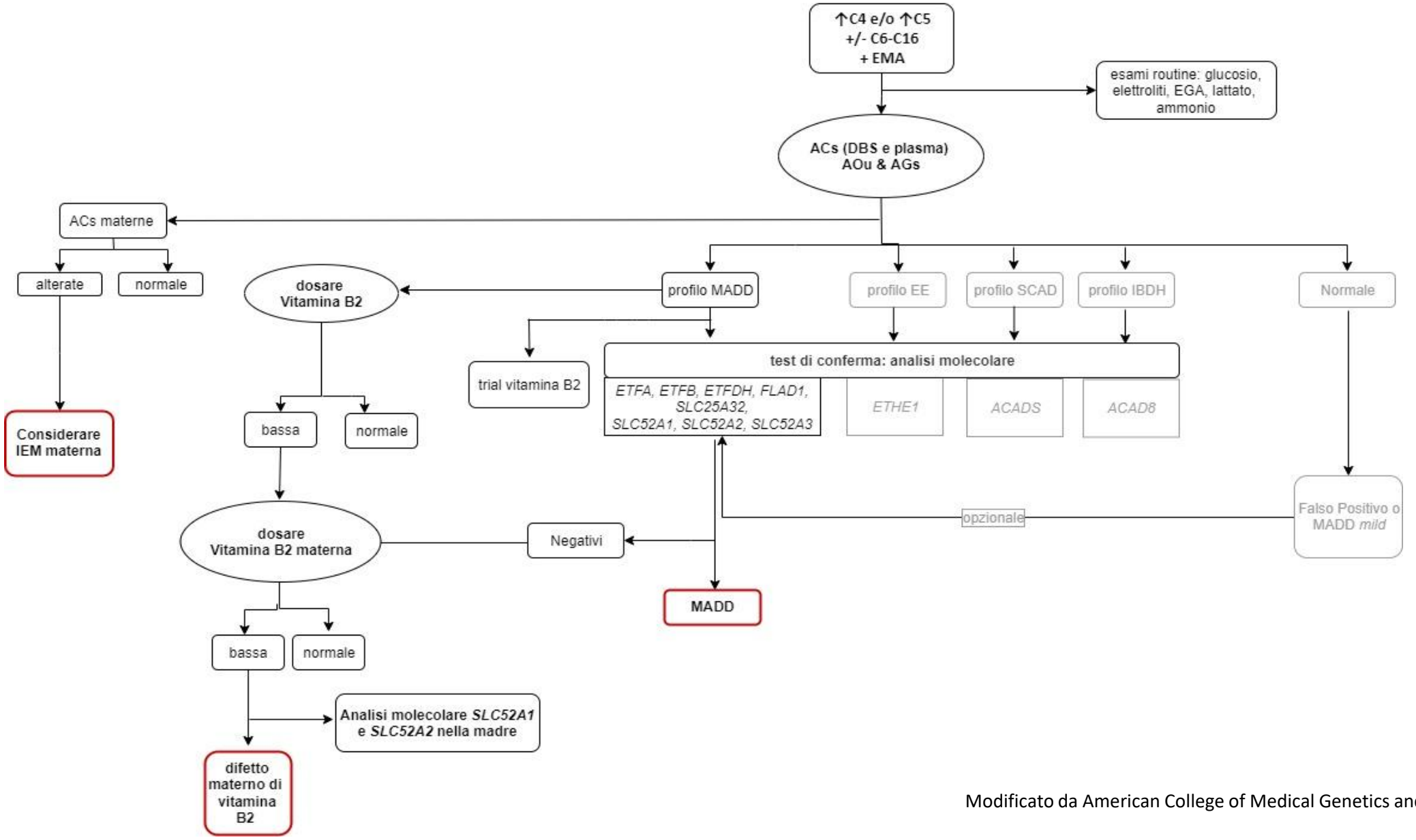
MGM Reports

Maternal vitamin deficiency mimicking multiple acyl-CoA dehydrogenase deficiency on newborn screening

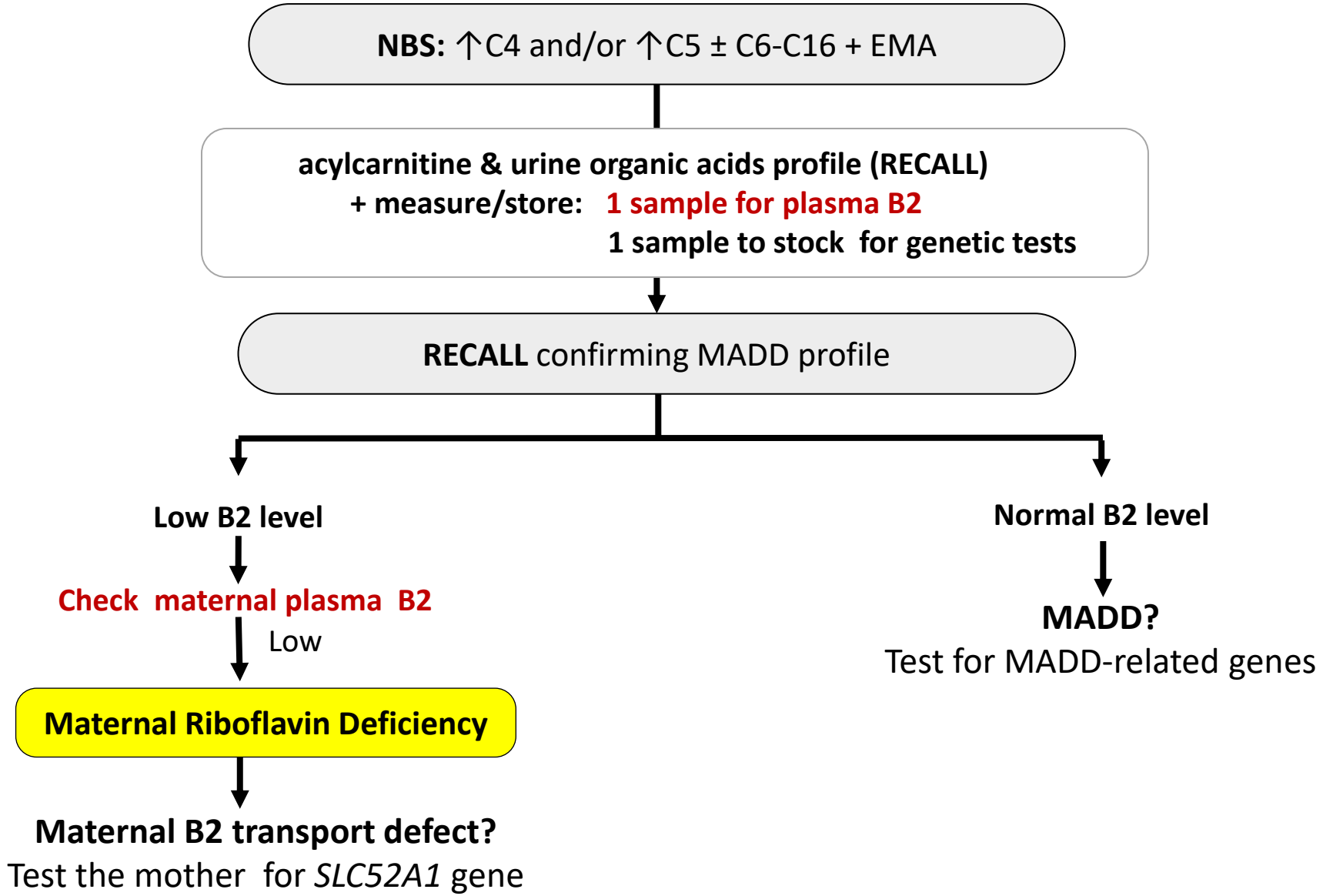
Gwendolyn Gramer[a,*], Georg F. Hoffmann[a], Julia B. Hennermann[b]

Proposal Algorithm for diagnosis implementation



Modificato da American College of Medical Genetics and Genomics

Proposal Algorithm for diagnosis implementation



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