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Introduzione

Lo screening neonatale esteso (SNE) si basa sul dosaggio in spettrometria di massa tandem (TMS) delle acilcarnitine (AC) e degli amminoacidi (AA) presenti in una goccia di sangue essiccato su carta (DBS). Per la conferma diagnostica e il follow-up dei pazienti possono essere utilizzate come matrici biologiche sia il DBS e sia il plasma.

Metodi & Pazienti

Sono state quantificate le AC e gli AA nel DBS e plasma di 270 soggetti sani italiani di differenti età (1-6 mesi, 6-12 mesi, 1-6 anni, 6-18 anni e >18 anni) al fine di ottenere i valori di riferimento e valutare le differenze quantitative tra DBS e plasma. Le acilcarnitine e gli amminoacidi sono estratti da DBS e plasma in accordo con il metodo Chromsystems¹ e butilati prima di essere analizzati in TMS. Le modalità di acquisizione TMS usate sono: il precursor ion per le acilcarnitine; Neutral Loss e MRM per gli amminoacidi. Le funzioni statistiche utilizzate per ogni analita nei differenti gruppi di età sono state: percentili, min, max, media, mediana, DS, T-test.

Risultati

Le Tabelle 1A, 1B, 1C, 1D mostrano i valori della media, minimo e massimo delle AC e delle AA in DBS e plasma nei 5 gruppi di età.

Nella Figura 1 sono rappresentati schematicamente gli andamenti all'aumentare dell'età delle AC e degli AA.

Nella Figura 2 sono rappresentate schematicamente le differenze quantitative delle AC e degli AA nelle matrici DBS e plasma.

Tabella 1A

Anionic	Site	EPR					PES				
		1-6	6-12	14-18	18-24	24-30	1-6	6-12	14-18	18-24	24-30
A1	Meta	31.72	28.81	23.66	14.94	26.30	50.27	43.67	36.37	30.17	37.07
	Para	31.72	28.81	23.66	14.94	26.30	50.27	43.67	36.37	30.17	37.07
	Ortho	31.72	28.81	23.66	14.94	26.30	50.27	43.67	36.37	30.17	37.07
C1	Meta	30.66	28.06	27.44	18.61	26.01	48.06	37.56	35.06	33.56	35.06
	Para	30.66	28.06	27.44	18.61	26.01	48.06	37.56	35.06	33.56	35.06
	Ortho	30.66	28.06	27.44	18.61	26.01	48.06	37.56	35.06	33.56	35.06
C2	Meta	18.61	18.61	17.91	17.91	17.91	43.56	43.56	43.56	43.56	43.56
	Para	18.61	18.61	17.91	17.91	17.91	43.56	43.56	43.56	43.56	43.56
	Ortho	18.61	18.61	17.91	17.91	17.91	43.56	43.56	43.56	43.56	43.56
C3	Meta	18.61	18.61	17.91	17.91	17.91	43.56	43.56	43.56	43.56	43.56
	Para	18.61	18.61	17.91	17.91	17.91	43.56	43.56	43.56	43.56	43.56
	Ortho	18.61	18.61	17.91	17.91	17.91	43.56	43.56	43.56	43.56	43.56
C4	Meta	18.61	18.61	17.91	17.91	17.91	43.56	43.56	43.56	43.56	43.56
	Para	18.61	18.61	17.91	17.91	17.91	43.56	43.56	43.56	43.56	43.56
	Ortho	18.61	18.61	17.91	17.91	17.91	43.56	43.56	43.56	43.56	43.56
C5	Meta	0.109	0.087	0.044	0.024	0.014	0.172	0.152	0.149	0.151	0.152
	Para	0.109	0.087	0.044	0.024	0.014	0.172	0.152	0.149	0.151	0.152
	Ortho	0.109	0.087	0.044	0.024	0.014	0.172	0.152	0.149	0.151	0.152
C6	Meta	0.109	0.087	0.044	0.024	0.014	0.172	0.152	0.149	0.151	0.152
	Para	0.109	0.087	0.044	0.024	0.014	0.172	0.152	0.149	0.151	0.152
	Ortho	0.109	0.087	0.044	0.024	0.014	0.172	0.152	0.149	0.151	0.152
C7	Meta	0.109	0.087	0.044	0.024	0.014	0.172	0.152	0.149	0.151	0.152
	Para	0.109	0.087	0.044	0.024	0.014	0.172	0.152	0.149	0.151	0.152
	Ortho	0.109	0.087	0.044	0.024	0.014	0.172	0.152	0.149	0.151	0.152
C8	Meta	0.109	0.087	0.044	0.024	0.014	0.172	0.152	0.149	0.151	0.152
	Para	0.109	0.087	0.044	0.024	0.014	0.172	0.152	0.149	0.151	0.152
	Ortho	0.109	0.087	0.044	0.024	0.014	0.172	0.152	0.149	0.151	0.152
C9	Meta	0.109	0.087	0.044	0.024	0.014	0.172	0.152	0.149	0.151	0.152
	Para	0.109	0.087	0.044	0.024	0.014	0.172	0.152	0.149	0.151	0.152
	Ortho	0.109	0.087	0.044	0.024	0.014	0.172	0.152	0.149	0.151	0.152
C10	Meta	0.109	0.087	0.044	0.024	0.014	0.172	0.152	0.149	0.151	0.152

Tabella 1B

Asset		Alpha					Pima				
		1-6	6-12	12-18	18-24	24-30	1-6	6-12	12-18	18-24	24-30
COC	Med	0.098	0.052	0.047	0.048	0.058	0.075	0.047	0.048	0.075	0.047
	Min	0.098	0.052	0.047	0.048	0.058	0.075	0.047	0.048	0.075	0.047
	Max	0.098	0.052	0.047	0.048	0.058	0.075	0.047	0.048	0.075	0.047
C12	Med	0.013	0.018	0.018	0.017	0.017	0.020	0.020	0.018	0.018	0.018
	Min	0.013	0.018	0.018	0.017	0.017	0.020	0.020	0.018	0.018	0.018
	Max	0.013	0.018	0.018	0.017	0.017	0.020	0.020	0.018	0.018	0.018
C18	Med	0.011	0.054	0.047	0.048	0.050	0.062	0.062	0.062	0.062	0.062
	Min	0.011	0.054	0.047	0.048	0.050	0.062	0.062	0.062	0.062	0.062
	Max	0.011	0.054	0.047	0.048	0.050	0.062	0.062	0.062	0.062	0.062
C3	Med	0.014	0.090	0.096	0.093	0.094	0.132	0.132	0.132	0.132	0.132
	Min	0.014	0.090	0.096	0.093	0.094	0.132	0.132	0.132	0.132	0.132
	Max	0.014	0.090	0.096	0.093	0.094	0.132	0.132	0.132	0.132	0.132
COC	Med	0.007	0.009	0.009	0.009	0.010	0.009	0.009	0.009	0.009	0.009
	Min	0.007	0.009	0.009	0.009	0.010	0.009	0.009	0.009	0.009	0.009
	Max	0.007	0.009	0.009	0.009	0.010	0.009	0.009	0.009	0.009	0.009
C12	Med	0.019	0.017	0.017	0.019	0.019	0.016	0.016	0.017	0.016	0.017
	Min	0.019	0.017	0.017	0.019	0.019	0.016	0.016	0.017	0.016	0.017
	Max	0.019	0.017	0.017	0.019	0.019	0.016	0.016	0.017	0.016	0.017
C18	Med	0.009	0.028	0.028	0.028	0.029	0.029	0.029	0.029	0.029	0.029
	Min	0.009	0.028	0.028	0.028	0.029	0.029	0.029	0.029	0.029	0.029
	Max	0.009	0.028	0.028	0.028	0.029	0.029	0.029	0.029	0.029	0.029
C3	Med	0.007	0.008	0.008	0.008	0.009	0.009	0.009	0.009	0.009	0.009
	Min	0.007	0.008	0.008	0.008	0.009	0.009	0.009	0.009	0.009	0.009
	Max	0.007	0.008	0.008	0.008	0.009	0.009	0.009	0.009	0.009	0.009
C12	Med	0.019	0.018	0.018	0.017	0.017	0.016	0.016	0.017	0.016	0.017
	Min	0.019	0.018	0.018	0.017	0.017	0.016	0.016	0.017	0.016	0.017
	Max	0.019	0.018	0.018	0.017	0.017	0.016	0.016	0.017	0.016	0.017
C18	Med	0.009	0.028	0.028	0.028	0.029	0.029	0.029	0.029	0.029	0.029
	Min	0.009	0.028	0.028	0.028	0.029	0.029	0.029	0.029	0.029	0.029
	Max	0.009	0.028	0.028	0.028	0.029	0.029	0.029	0.029	0.029	0.029
C3	Med	0.009	0.006	0.005	0.006	0.007	0.010	0.010	0.009	0.010	0.009
	Min	0.009	0.006	0.005	0.006	0.007	0.010	0.010	0.009	0.010	0.009
	Max	0.009	0.006	0.005	0.006	0.007	0.010	0.010	0.009	0.010	0.009
COC	Med	0.014	0.023	0.039	0.034	0.033	0.039	0.039	0.039	0.039	0.039
	Min	0.014	0.023	0.039	0.034	0.033	0.039	0.039	0.039	0.039	0.039
	Max	0.014	0.023	0.039	0.034	0.033	0.039	0.039	0.039	0.039	0.039
C12	Med	0.007	0.007	0.006	0.007	0.007	0.006	0.006	0.007	0.006	0.007
	Min	0.007	0.007	0.006	0.007	0.007	0.006	0.006	0.007	0.006	0.007
	Max	0.007	0.007	0.006	0.007	0.007	0.006	0.006	0.007	0.006	0.007
C18	Med	0.009	0.036	0.036	0.036	0.037	0.037	0.037	0.037	0.037	0.037
	Min	0.009	0.036	0.036	0.036	0.037	0.037	0.037	0.037	0.037	0.037
	Max	0.009	0.036	0.036	0.036	0.037	0.037	0.037	0.037	0.037	0.037
C3	Med	0.009	0.014	0.023	0.034	0.033	0.039	0.039	0.039	0.039	0.039
	Min	0.009	0.014	0.023	0.034	0.033	0.039	0.039	0.039	0.039	0.039
	Max	0.009	0.014	0.023	0.034	0.033	0.039	0.039	0.039	0.039	0.039
COC	Med	0.013	0.013	0.006	0.007	0.007	0.014	0.006	0.006	0.014	0.006
	Min	0.013	0.013	0.006	0.007	0.007	0.014	0.006	0.006	0.014	0.006
	Max	0.013	0.013	0.006	0.007	0.007	0.014	0.006	0.006	0.014	0.006
C12	Med	0.009	0.018	0.018	0.018	0.019	0.021	0.021	0.018	0.021	0.018
	Min	0.009	0.018	0.018	0.018	0.019	0.021	0.021	0.018	0.021	0.018
	Max	0.009	0.018	0.018	0.018	0.019	0.021	0.021	0.018	0.021	0.018
C18	Med	0.009	0.028	0.028	0.028	0.029	0.029	0.029	0.029	0.029	0.029
	Min	0.009	0.028	0.028	0.028	0.029	0.029	0.029	0.029	0.029	0.029
	Max	0.009	0.028	0.028	0.028	0.029	0.029	0.029	0.029	0.029	0.029
C3	Med	0.009	0.006	0.006	0.006	0.007	0.009	0.009	0.009	0.009	0.009
	Min	0.009	0.006	0.006	0.006	0.007	0.009	0.009	0.009	0.009	0.009
	Max	0.009	0.006	0.006	0.006	0.007	0.009	0.009	0.009	0.009	0.009
COC	Med	0.007	0.008	0.008	0.008	0.009	0.009	0.009	0.009	0.009	0.009
	Min	0.007	0.008	0.008	0.008	0.009	0.009	0.009	0.009	0.009	0.009
	Max	0.007	0.008	0.008	0.008	0.009	0.009	0.009	0.009	0.009	0.009
C12	Med	0.019	0.018	0.018	0.017	0.017	0.016	0.016	0.017	0.016	0.017
	Min	0.019	0.018	0.018	0.017	0.017	0.016	0.016	0.017	0.016	0.017
	Max	0.019	0.018	0.018	0.017	0.017	0.016	0.016	0.017	0.016	0.017
C18	Med	0.009	0.028	0.028	0.028	0.029	0.029	0.029	0.029	0.029	0.029
	Min	0.009	0.028	0.028	0.028	0.029	0.029	0.029	0.029	0.029	0.029
	Max	0.009	0.028	0.028	0.028	0.029	0.029	0.029	0.029	0.029	0.029
C3	Med	0.009	0.014	0.023	0.034	0.033	0.039	0.039	0.039	0.039	0.039
	Min	0.009	0.014	0.023	0.034	0.033	0.039	0.039	0.039	0.039	0.039
	Max	0.009	0.014	0.023	0.034	0.033	0.039	0.039	0.039	0.039	0.039
COC	Med	0.007	0.009	0.009	0.009	0.010	0.009	0.009	0.009	0.009	0.009
	Min	0.007	0.009	0.009	0.009	0.010	0.009	0.009	0.009	0.009	0.009
	Max	0.007	0.009	0.009	0.009	0.010	0.009	0.009	0.009	0.009	0.009
C12	Med	0.019	0.017	0.017	0.019	0.019	0.016	0.016	0.017	0.016	0.017
	Min	0.019	0.017	0.017	0.019	0.019	0.016	0.016	0.017	0.016	0.017
	Max	0.019	0.017	0.017	0.019	0.019	0.016	0.016	0.017	0.016	0.017
C18	Med	0.009	0.028	0.028	0.028	0.029	0.029	0.029	0.029	0.029	0.029
	Min	0.009	0.028	0.028	0.028	0.029	0.029	0.029	0.029	0.029	0.029
	Max	0.009	0.028	0.028	0.028	0.029	0.029	0.029	0.029	0.029	0.029
C3	Med	0.009	0.006	0.006	0.006	0.007	0.009	0.009	0.009	0.009	0.009
	Min	0.009	0.006	0.006	0.006	0.007	0.009	0.009	0.009	0.009	0.009
	Max	0.009	0.006	0.006	0.006	0.007	0.009	0.009	0.009	0.009	0.009

Tabella 1C

Analysis	SBS						Peters					
	1.4	6.12	1.4	6.12	1.4	6.12	1.4	6.12	1.4	6.12		
CBI	Mean	0.915	0.845	0.91	0.849	0.849	0.91	0.839	0.913	0.881	0.91	
	Std	0.008	0.008	0.008	0.008	0.008	0.008	0.008	0.008	0.008	0.008	
CBI1	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI2	Mean	0.922	0.912	0.921	0.912	0.913	0.921	0.913	0.926	0.916	0.922	
	Std	0.007	0.007	0.007	0.007	0.007	0.007	0.007	0.007	0.007	0.007	
CBI3	Mean	0.951	0.941	0.951	0.941	0.941	0.951	0.941	0.951	0.941	0.951	
	Std	0.005	0.005	0.005	0.005	0.005	0.005	0.005	0.005	0.005	0.005	
CBI4	Mean	0.978	0.978	0.978	0.978	0.978	0.978	0.978	0.978	0.978	0.978	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI5	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI6	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI7	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI8	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI9	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI10	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI11	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI12	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI13	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI14	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI15	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI16	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI17	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI18	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI19	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI20	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI21	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI22	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI23	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI24	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI25	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI26	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI27	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI28	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI29	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI30	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI31	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI32	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI33	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI34	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI35	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI36	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI37	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI38	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI39	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI40	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI41	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI42	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI43	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI44	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI45	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI46	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI47	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI48	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI49	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI50	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI51	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001	
CBI52	Mean	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	0.988	
	Std	0.001	0.001	0.001	0.001	0.001	0.001	0.001	0.001			

Tabella 1D

[illegible]

Figura 1: Andamento con l'età delle AC e degli AA in DBS e Plasma

Analiti	Andamento con età	
	DBS	Plasma
C0		
C2		
C3		
C4		
C5:1		
C5		
C4-OH		
C6		
C5-OH		
C8:1		
C8		
C3DC		
C10:1		
C10		
C4DC		
C5DC		
C12:2		
C12:1		
C12		
C6DC		
C12-OH		
C14:2		
C14:1		
C14		
C14:1-OH		
C14-OH		
C16:1		
C16		
C16:1-OH		
C16-OH		
C18:2		
C18:1		
C18		
C18:2-OH		
C18:1-OH		
C18-OH		
C16DC		

Anali	Andamento con l'età	
	Inf.	Ad.

	D85	Plasma
GLY		
ALA		
VAL		
LEU		
MET		
PHE		
TYR		
ASP		
GLU		
CYS		
ARG		
CYS		
PRO		
HOMOCYS		
ARG/SUCC-1		

-  Andamento decrescente con l'età
-  Andamento crescente con l'età
-  Andamento costante con l'età
-  Andamento crescente e poi decrescente con l'età
-  Andamento decrescente e poi crescente con l'età

Figura 2: Differenze quantitative delle AC e degli AA in DBS e Plasma

Analyte	DBS	Plasma
C0		
C2		
C3		
C4		
C5-1		
C5		
C4-OH		
C6		
C5-OH		
C8-1		
C8		
C9OC		
C10-1		
C10		
C9OC		
C12-2		
C12-1		
C12		
C9OC		
C12-OH		
C14-2		
C14-1		
C14		
C14-1-OH		
C14-OH		
C16-1		
C16		
C16-1-OH		
C18-OH		
C18-2		
C18-1		
C18		
C18-2-OH		
C18-1-OH		
C18-OH		
C18OC		
C18OC		

Analiiti	DBS	Plasma
CRP		

GLY		
ALA		
VAL		
LEU		
MET		
PHE		
TYR		
ASP	6-18 A > 18 A	1-6 M; 6-12 M; 1-6 A
GLU		
ORN		
ARG		
OTR		
PRO		
HOMOCYTR		
ARGSUCC		

Valori più aumentati

Conclusioni

I dati quantitativi delle AC e degli AA nel DBS e plasma, correlate con l'età, aggiungono nuove importanti informazioni che permettono una migliore identificazione degli errori congeniti del metabolismo ad esordio tardivo ed un corretto approccio terapeutico e follow-up dei pazienti nel tempo.

Bibliografia

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