

Austrian Newbornscreening

- from PKU (1966) to SMA (2021) -

the new experience !

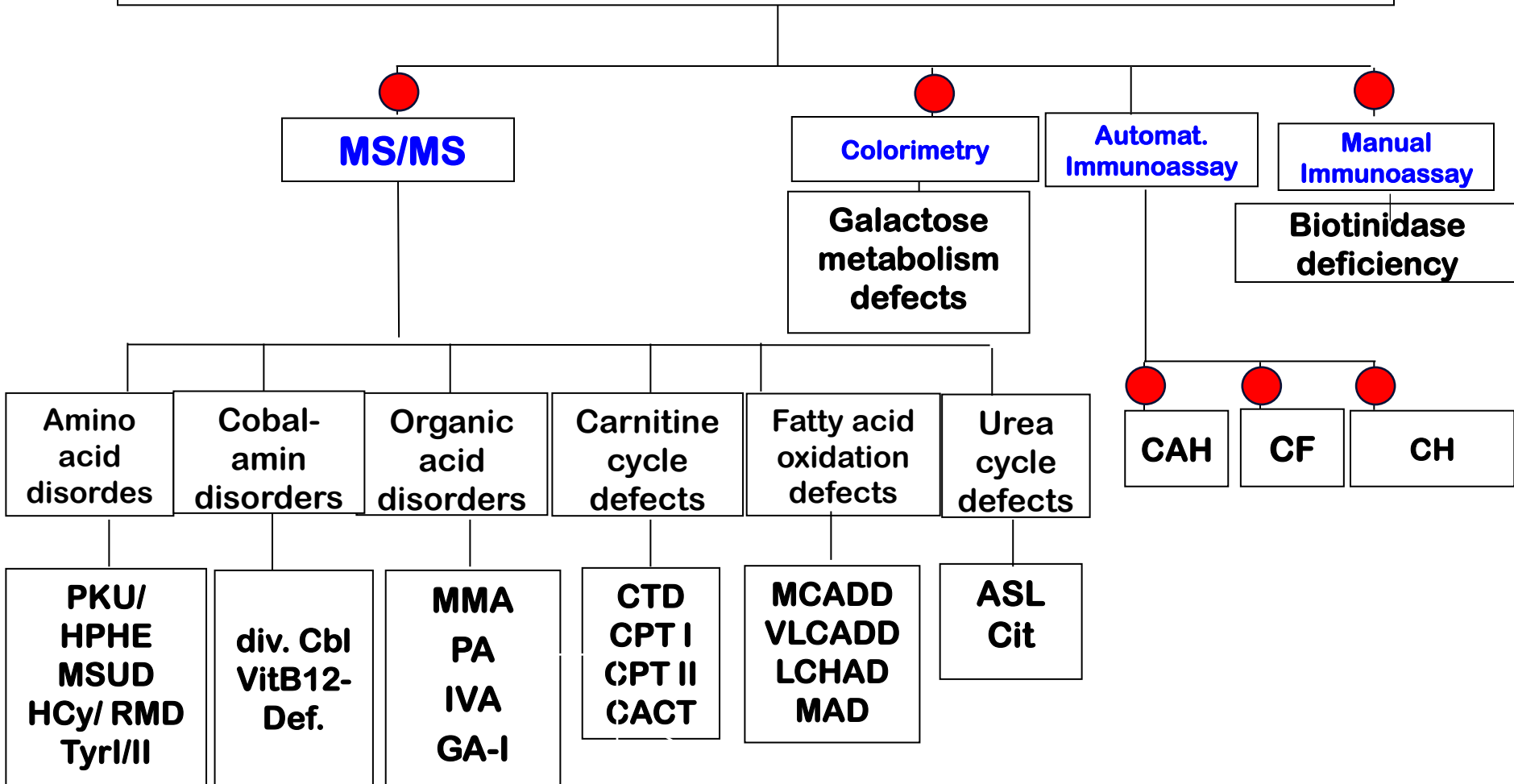
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&

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Austrian Newborn Screening



Country	ch	cah	cf	pkuhpa	msud	hcl	tyrl	asa	citl	arg	hptl_III	tyrl_III	citll	gal	hva	mmachl	pa	3mcc	gal	hscd	3img	bkt	mma	mcadd	vicadd	lchadd	cpfl	cpfl	cud	scadd	schadd	decr	s-s	btha	sc	bio	gal	udp	SCID	CCHD	N disorders for		
Albania	x			x																																					2		
Austria	x	x	x	x	x	x	x	x	x	x	x			x	x	x	x	x	x	x	x		x	x	x	x	x	x	x							x	x	x				28	
Azerbaijan																																											
Belarus	x			x																																						2	
Belgium (Flemish)	x	x		x	x									x	x	x	x		x																							11	
Belgium (French)	x	x ²	x ²	x	x	x	x					x		x	x	x	x		x														x ²	x ²	x ²	x ²	x					15	
Bosnia-Herzegovina	x	x		x																																						3	
Bulgaria	x	x		x																																						3	
Croatia	x			x																																						2	
Cyprus	x			x																																						2	
Czech Republic	x	x	x	x	x									x	x																											12	
Denmark	x	x		x	x									x	x																											16	
Estonia	x			x	x	x	x	x	x						x	x	x	x																								2	
Finland	x																																									1	
France	x	x	x	x																																						5	
Georgia																																											
Germany	x	x	x	x	x									x	x																											16	
Greece	x			x																																						3	
Hungary	x			x	x	x	x	x	x																																	26	
Iceland	x			x	x									x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x										26		
Ireland	x		x	x	x	x																																				6	
Italy (*)	x	x	x ²	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x										33		
% coverage	100	42	81	100	48	30	48	30	30	30	30	30	30	30	30	30	30	30	30	30	30	30	30	30	30	30	30	30	30	30	30	0.8											
N. labs	25	4	15	19	11	8	9	8	8	8	8	8	8	8	8	8	8	8	8	8	8	8	8	8	8	8	8	8	8	8	8												
Kazakhstan	x			x																																						2	
Kosovo																																											0
Latvia	x			x																																						2	
Lithuania	x			x																																						2	
Luxembourg	x	x		x																																						4	
Macedonia	x																																									1	
Malta	x																																									3	
Moldova																																											
Montenegro	x																																									1	
Netherlands	x	x	x	x	x	x	x							x	x																											20	
Norway	x	x	x	x	x	x	x							x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x												22	
Poland	x	x	x	x	x	x	x							x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x												21	
Portugal	x			x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x												25	
Romania	x			x																																						2	
Russia	x	x	x	x																																						5	
Serbia	x			x																																						2	
Slovakia	x	x	x	x																																						4	
Slovenia	x			x																																						2	
Spain ⁴ ^{1/10/2016}	x	x	x	x	x	x	x	x	x	x				x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x												27		
Sweden	x	x		x	x	x	x							x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x													24	
Switzerland ¹	x	x	x	x	x ²									x ²																											9		
Turkey	x			x																																						3	
Turkmenistan	x	x	x	x																																						4	
Ukraine																																											
United Kingdom	x		x	x	x	x								x	x																											12	
Uzbekistan	x		x	x																																						3	

Source: G. Loeber, ISNS. 2017 unpublished

Newborn screening for SMA: the Austrian experience

Pilot project: Procedures

- 06/2021 – 07/2022
- ~ 90.000 newborns
- Using a commercial kit* from Perkin Elmer (simultaneously TREC and KREC for severe primary immunodeficiencies are analyzed)
- In case of positive result -> repeat from same sample -> if positive (**was always**) -> information of designated neuropediatric experts from center for the particular region, who contact parents and initiate diagnosis and treatment

*real time pcr

Newborn screening for SMA: the Austrian experience

Pilot project: Preliminary Results

- Approx. 60.000 newborns analyzed
- Positive newborns: 11
- 2 copies: 4/11 // 3 copies: 1/11 // 4 copies: 4/11 // 5 copies: 1/11
- 1/11 false positive (undescribed "missense" variant (c.842G>C))
- Gene therapy: 5/10 (50%) – all without symptoms at recall !!!
- Time till gene therapy: within 21 days