

Table S1. Statements of the German Guideline for Primary Congenital Hypothyroidism (CH) which were not further examined in the present manuscript [1].

Statement 5	<ul style="list-style-type: none"> • For goiter: sequencing of the TPO gene • With a simultaneous cleft palate: exclusion of FOXE1 gene mutation • For coexistent movement disorders: exclusion of NKX2.1 gene mutation
Statement 6	<ul style="list-style-type: none"> • Sole L-T₄ supplementation is recommended as medication of choice
Statement 10	<p>Timing of reevaluation of thyroid function:</p> <ul style="list-style-type: none"> • In case of iodine exposure: after 3 months of therapy • In case of decreasing previously detected auto-antibody titers or stable blood TSH or if thyroid gland is detectable: after the 2nd year of life

Table S2. Additional data on German newborn screening [2].

Year	Number of confirmed cases of CH in newborn screening	Prevalence	Recall rate [%]	Positive predictive value PPV [%]	Specificity [%]
2004	222	1:3,270	0.13	22.67	
2005	187	1:3,728	0.24	11.15	99.79
2006	165	1:4,156	0.17	14.49	99.86
2007	163	1:4,202	0.14	16.98	99.88
2008	184	1:3,709	0.09*	28.62*	99.94*
2009	195	1:3,441	0.09*	29.39*	99.88*
2010	207	1:3,275	0.08*	35.23*	
2011	207	1:3,201	0.08*	35.11*	
2012	205	1:3,285	0.13		
2013	211	1:3,233	0.12		
2014	213	1:3,356	0.14		
2015	235	1:3,139	0.13		
2016	242	1:3,273	0.12		
2017	279	1:2,813	0.13	28.01	99.91

*data derived only from tests performed \geq 36 hours.

Table S3. Morphology of thyroid gland, examined via sonography, divided before and after guideline appearance (groups A and B).

	Group A "before guideline" 10/ 2001 – 1/2011	Group B "after guideline" 2/2011 – 05/2020
Thyroid gland in loco typico	61.9 (n=84)	62.7 (n=118)
Ectopia	7.4 (n=54)	6.7 (n=90)
Athyreosis	24.1 (n=58)	38.5 (n=96)

Results are given in %. N is the total number of patients available for each analysis as explained in the results part of the main article (page 5).

Table S4. Genetic findings indicating a) dysgenesis or b) dyshormonogenesis, c) no mutation found, divided before and after guideline appearance (groups A and B).

	Group A "before guideline" 10/ 2001 – 1/2011	Group B "after guideline" 2/2011 – 05/2020
a) Dysgenesis		
NKX2.1/TTF1-gene mutation	9.1 (n=11)	0.0 (n=13)
PAX8-gene mutation	9.1 (n=11)	7.7 (n=13)
TSH-receptor-gene mutation	9.1 (n=11)	15.4 (n=13)
b) Dyshormonogenesis		
SLC26A4 (pendrin)-gene mutation	18.2 (n=11)	0.0 (n=13)
Thyroid peroxidase (TPO)-gene mutation	18.2 (n=11)	38.5 (n=13)
Thyreoglobuline (Tg) gene mutation	18.2 (n=11)	0.0 (n=13)
c) No mutation found		
No mutation found	18.2 (n=11)	38.5 (n=13)

Results are given in %. N is the total number of patients available for each analysis as explained in the results part of the main article (page 5).

1. H, K. Diagnostik, Therapie und Verlaufskontrolle der Primären angeborenen Hypothyreose. Available online: https://www.awmf.org/uploads/tx_szleitlinien/027-0171_S2k_Primaere_Aangeborene_Hypothyreose_2011-abgelaufen.pdf (accessed on 30.01.2021).
2. e.V., D.G.f.r.N. DGNS Screeningreports 2004-2017. Available online: <http://www.screening-dgns.de/reports.php> (accessed on 29.01.2021)