

Supplementary Table 4 Up-to-date performance of newborn screening programs in different countries: a survey combined a literature review.

Country of the NBS program	Age at time of initial screening	The percent of specimens deemed unsatisfactory for analysis	Period between sample collection and receipt by the lab	Period between receipt by the lab and reporting the results	N of abnormal initial screening	N of recalled cases	N of confirmed cases	Recall rate	PPV
Germany	51.94% withing 36-48 hours; 43.22% within 48-72 hours.	0.45%	71.28% in 3 days	75.1% of the results are reported within 24 hours.	21195 (not limited to PKU/HPA and CH)	17864 (302 for PKU/HPA; 922 for CH)	157 for PKU/HPA; 246 for CH	84.28% (not limited to PKU/HPA and CH).	51.99% for PKU/HPA; 26.68% for CH.
UK	95.6% on days 5 to 8; 2.1% on or before day 4; 77.9% on day 5; 2.3% on or after day 9.	3.22%	92.3% of all samples received within 3 working days. 97.0% within 4 working days	97.1% of negative results letters are despatched direct to parents by 6 weeks of age	1.37 per 10,000 babies (107/779,688) for PKU	/	/	/	/

China	The time is required to be after 72 hours of birth but within 7 days, and full breastfeeding before collection.	0.27% (median: 0.13%, IQR: 0.05%-0.34%)	94% in 5 days (median: 99.56%)	97.50% within 5 days (median: 100%, IQR: 98.48%-100%)	6292 for PKU/HPA; 34203 for CH.	4974 for PKU/HPA; 31958 for CH.	303 for PKU/HPA; 2154 for CH.	79.05% for PKU/HPA; 93.44% for CH.	6.09% for PKU/HPA; 6.74% for CH.
America	> 90% within 48 h (median: 97%)	1.5% (median)	28.9% - 56.5% in one day (median: 41.8%), 70.3% - 88.45% in two days (median: 81%).	88.3%-99.3% within 4 days (median: 94.2%)	\	\	\	\	\

NBS, newborn screening; PPV, predictive positive rate; PKU/HPA, phenylketonuria / hyperphenylalaninemia; CH, congenital hypothyroidism; IQR: inter quartile range.