

ORIGINAL ARTICLE

Results of a Pilot External Quality Assessment Scheme for Genetic Testing of Newborns with Spinal Muscular Atrophy

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SUMMARY

Background: This study aims to evaluate the ability of laboratories to perform spinal muscular atrophy (SMA) genetic testing in newborns based on dried blood spot (DBS) samples, and to provide reference data and advance preparation for establishing the pilot external quality assessment (EQA) scheme for SMA genetic testing of newborns in China.

Methods: The pilot EQA scheme contents and evaluation principles of this project were designed by National Center for Clinical Laboratories (NCCL), National Health Commission. Two surveys were carried out in 2022, and 5 batches of blood spots were submitted to the participating laboratory each time. All participating laboratories conducted testing upon receiving samples, and test results were submitted to NCCL within the specified date.

Results: The return rates were 75.0% (21/28) and 95.2% (20/21) in the first and second surveys, respectively. The total return rate of the two examinations was 83.7% (41/49). Nineteen laboratories (19/21, 90.5%) had a full score passing on the first survey, while in the second survey twenty laboratories (20/20, 100%) scored full.

Conclusions: This pilot EQA survey provides a preliminary understanding of the capability of SMA genetic testing for newborns across laboratories in China. A few laboratories had technical or operational problems in testing. It is, therefore, of importance to strengthen laboratory management and to improve testing capacity for the establishment of a national EQA scheme for newborn SMA genetic testing.

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Supplementary Data

Table S1. Scores for the first external quality survey of newborn SMA genetic testing in 2022.

Lab ID	202211			202212			202213		202214		202215			Total
	SMN1	SMN2	Sub-total	SMN1	SMN2	Sub-total	SMN1	Sub-total	SMN2	Sub-total	SMN1	SMN2	Sub-total	
lab1	14	10	24	14	10	24	14	14	14	14	14	10	24	100
lab2	14	10	24	14	10	24	14	14	14	14	14	10	24	100
lab3	14	10	24	14	10	24	14	14	14	14	14	10	24	100
lab4	14	10	24	14	10	24	14	14	14	14	14	10	24	100
lab5	14	10	24	14	10	24	14	14	14	14	14	10	24	100
lab6	14	10	24	14	10	24	14	14	14	14	14	10	24	100
lab7	14	10	24	14	10	24	14	14	14	14	14	10	24	100
lab8	14	10	24	14	10	24	14	14	14	14	14	10	24	100
lab9	14	10	24	14	10	24	14	14	14	14	14	10	24	100
lab10	14	10	24	14	10	24	14	14	14	14	14	10	24	100
lab11	14	10	24	14	10	24	14	14	14	14	14	10	24	100
lab12	14	10	24	14	10	24	14	14	14	14	14	10	24	100
lab13	14	10	24	14	10	24	14	14	14	14	14	10	24	100
lab14	14	10	24	14	10	24	14	14	14	14	14	10	24	100
lab15	14	10	24	14	10	24	14	14	14	14	14	10	24	100
lab16	14	10	24	14	10	24	14	14	14	14	14	10	24	100
lab17	14	10	24	14	10	24	14	14	14	14	14	10	24	100
lab18	14	10	24	14	10	24	14	14	14	14	14	10	24	100
lab19	14	10	24	14	10	24	14	14	14	14	14	10	24	100
lab20	14	10	24	14	10	24	14	14	14	14	14	0	14	90
lab21	14	10	24	14	10	24	14	14	14	14	14	0	14	90

Table 2. Scores for the second external quality survey of newborn SMA genetic testing in 2022.

Lab ID	202221		202222		202223			202224			202225			Total
	SMN1	Sub-total	SMN1	Sub-total	SMN1	SMN2	Sub-total	SMN1	SMN2	Sub-total	SMN1	SMN2	Sub-total	
lab1	14	14	14	14	14	10	24	14	10	24	14	10	24	100
lab2	14	14	14	14	14	10	24	14	10	24	14	10	24	100
lab3	14	14	14	14	14	10	24	14	10	24	14	10	24	100
lab4	14	14	14	14	14	10	24	14	10	24	14	10	24	100
lab8	14	14	14	14	14	10	24	14	10	24	14	10	24	100
lab11	14	14	14	14	14	10	24	14	10	24	14	10	24	100
lab12	14	14	14	14	14	10	24	14	10	24	14	10	24	100
lab13	14	14	14	14	14	10	24	14	10	24	14	10	24	100
lab14	14	14	14	14	14	10	24	14	10	24	14	10	24	100
lab15	14	14	14	14	14	10	24	14	10	24	14	10	24	100
lab16	14	14	14	14	14	10	24	14	10	24	14	10	24	100
lab17	14	14	14	14	14	10	24	14	10	24	14	10	24	100
lab18	14	14	14	14	14	10	24	14	10	24	14	10	24	100
lab20	14	14	14	14	14	10	24	14	10	24	14	10	24	100
lab21	14	14	14	14	14	10	24	14	10	24	14	10	24	100
lab29	14	14	14	14	14	10	24	14	10	24	14	10	24	100
lab30	14	14	14	14	14	10	24	14	10	24	14	10	24	100
lab31	14	14	14	14	14	10	24	14	10	24	14	10	24	100
lab32	14	14	14	14	14	10	24	14	10	24	14	10	24	100
lab33	14	14	14	14	14	10	24	14	10	24	14	10	24	100

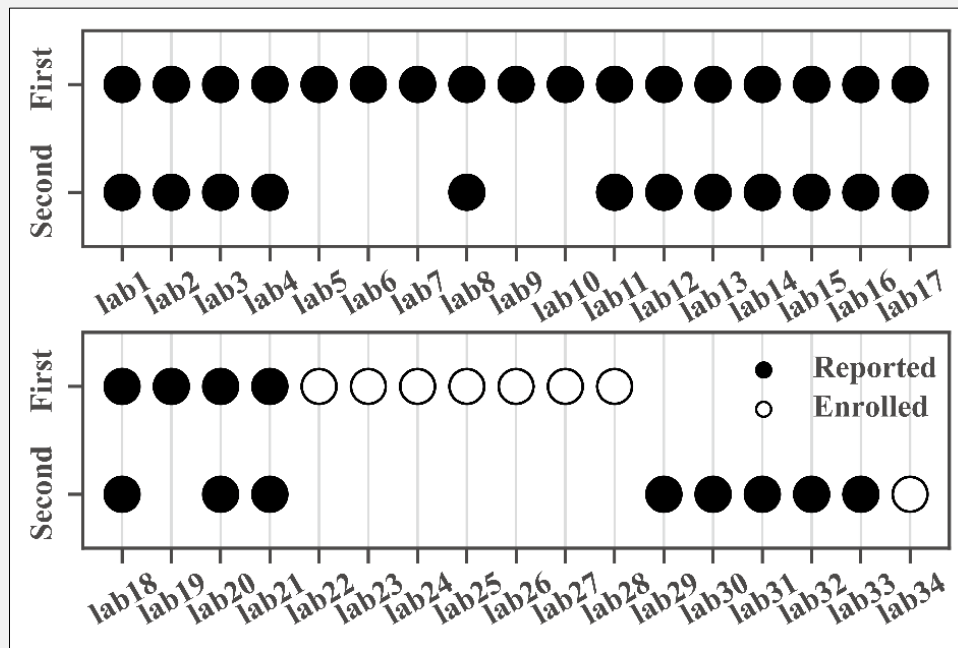


Figure 1. The participation of all 34 laboratories enrolled in the two surveys. Solid dots represent that the participant reported the results of copy number analysis, hollow dots represent enrollment only.