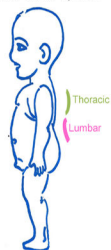


A Thoracic lumbar
spina bifida aperta



Total number of cases affected with only Thoracic lumbar spina bifida aperta	11
Associated pathway	Lipid metabolism
Occurrences in NTDs vs. in control	3/11 vs. 10/225
P value	0.029
Burden Gene(s)	<i>LEPR</i> (1), <i>APOB:APOB</i> (1)

B Thoracic
spina bifida aperta



Total number of cases affected with only Thoracic spina bifida aperta	2
Associated pathway	Protein processing
Occurrences in NTDs vs. in control	2/2 vs. 19/225
P value	0.037
Burden Gene(s)	<i>NUP98:NUP188</i> (1)

C Thoracic lumbar
spina bifida occulta



Total number of cases affected with only Thoracic lumbar spina bifida occulta	5
Associated pathway	Chromatin modification
Occurrences in NTDs vs. in control	4/5 vs. 27/225
P value	0.013
Burden Gene(s)	<i>CECR2</i> (2), <i>PRMT1</i> (1), <i>HIRA</i> (1)
Associated pathway	Neural development
Occurrences in NTDs vs. in control	3/5 vs. 12/225
P value	0.008
Burden Gene(s)	<i>ADNP</i> (1), <i>TRPM6</i> (2)

D Lumbar
spina bifida cystica



Total number of cases affected with only Lumbar spina bifida cystica	3
Associated pathway	Retinoid metabolism
Occurrences in NTDs vs. in control	1/3 vs. 0/225
P value	0.017
Burden Gene(s)	<i>CYP26B1</i>

E Thoracic
spina bifida cystica



Total number of cases affected with only Thoracic spina bifida cystica	1
Associated pathway	Apoptosis
Occurrences in NTDs vs. in control	1/1 vs. 4/225
P value	0.042
Burden Gene(s)	<i>CASP9</i>

Figure S1 Zou et al.