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Genotype call for chromosomal deletions using read-depth from whole-genome sequence variants in cattle

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Conclusions

- Genotype of a known deletion-locus could be inferred from the auxiliary read-depth data provided with whole-genome sequence variant calls
- ....could be used to extend reference panel for imputing deletions
- ....will facilitate inclusion of CNVs in GWAS and genomic prediction

Objective

- To estimate genotype likelihood of a ~3.3kb deletion on chromosome 21 from read-depth data

Acknowledgements

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Method

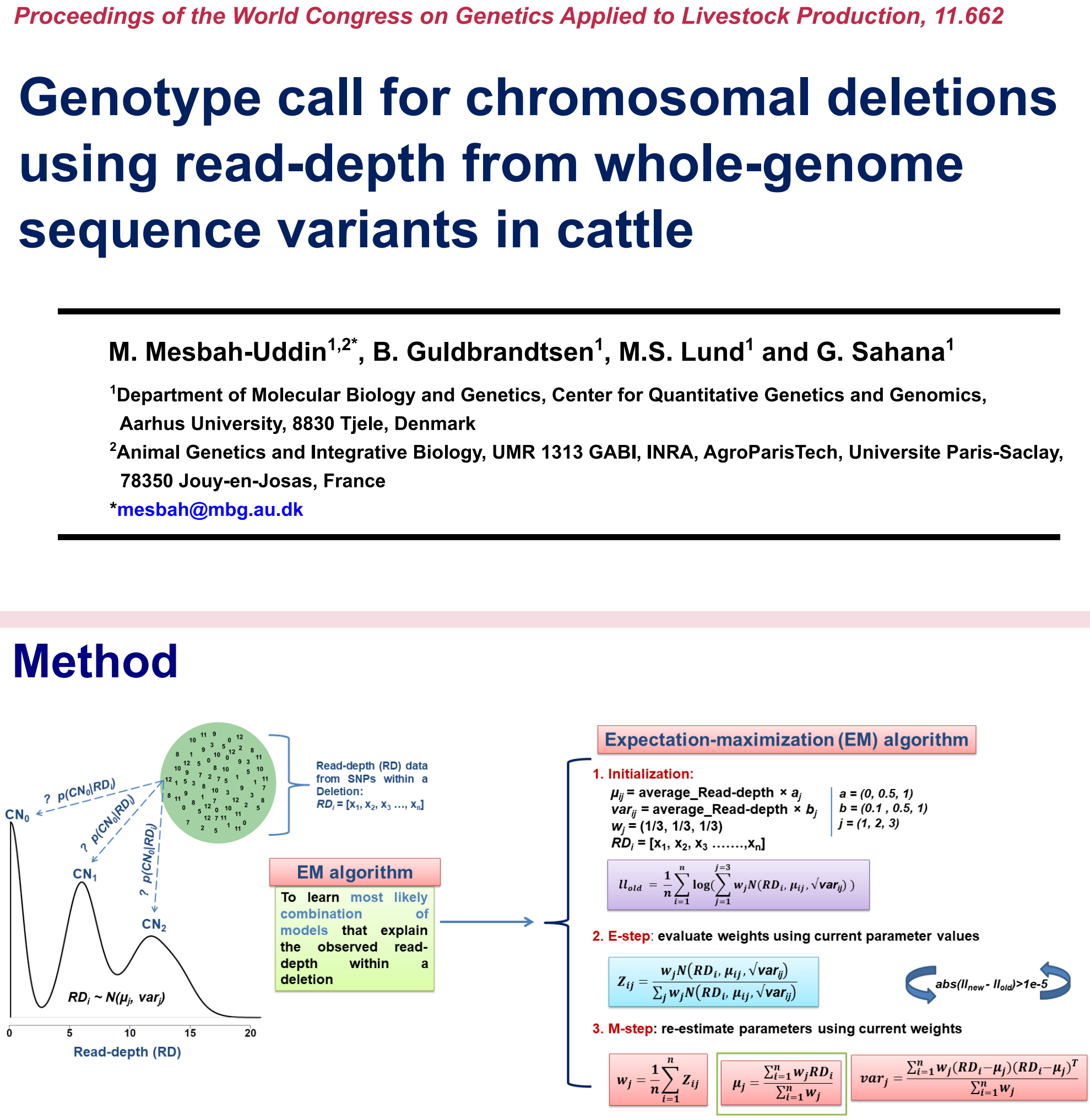

Figure 1. Graphical illustration of read-depth genotyping at a deletion locus using Gaussian mixture model.

Results

- 60 Holstein with known brachyspina status
- 13 carriers (BY)
- 47 non-carriers (TY)

		Actual class: BY	Actual class: TY
Predicted class	BY	13	3
	TY	0	44

Prediction accuracy = 95%

FDR = 18.8%

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