Validation of genomic and genetic evaluations

Esa Mäntysaari\textsuperscript{1}, Minna Koivula\textsuperscript{1},
Gert P. Aamand\textsuperscript{2}, and Ismo Strandén\textsuperscript{1}

\textsuperscript{1} Biometrical Genetics, Luke, Finland
\textsuperscript{2} NAV, Denmark
Validation of genetic/genomic evaluations

1. Development of genetic evaluations
   (Repeatability and bias of estimated breeding values for dairy bulls and bull dams calculated from animal model evaluations. Uimari & Mäntysaari; Animal Production 1993)

2. Continuous quality control for national evaluations

3. Monitor the reliability of EBVs as MACE input

4. Provide international (and EU) standards for GEBVs
Validation of genetic/genomic evaluations

Interbull validation tests I, II and III are all testing the genetic trend


– Actually, they are testing estimated environmental trend
– Tests rely on repeated records (daughters) for sires over time
– Will become unusable as soon as all sires are young
  • Need for alternative tests

GEBV validation test is cross validation test based on forward prediction

– Designed at the time when multi-step genomic evaluation was standard
GEBV validation test

Model I

\[ \text{DYD} = b_0 + b_1 \ \text{GEBV} + e \]

Model II

\[ \text{DYD} = b_0 + b_1 \ \text{EBV} + e \quad \% \quad \text{i.e. EBV is parent average} \]

- EBV and GEBV are estimated using truncated (-4 years) data
- DYD (or DRP) are estimated using full data

✓ Regression \( b_1 = 1.00 \)
if the evaluations and DYD are consistent

✓ GEBV should explain more of DYD than the EBV

GEBV validation test problems

• DYD (or DRP) for the estimation of GEBV (or PA) are from the full data
  – Most GEBV are based on MACE proofs, no truncated data MACE available

• Full data DYD or DRP are difficult to define when:
  – Test day models,
  – maternal effect models,
  – multi-lactation multi-trait
GEBV validation test problems (ssGBLUP)

- Generally badly suited for testing genomic animal models (single-step GBLUP)
  - Validation bulls should not have daughters, but the daughters might be essential part of genomic reference population
  - Validation can be done by predicting cow phenotypes (not cure to above)

- Generally young bulls highly selected according to GEBVs
  - Eventually the bull based validation $R^2$ start to decrease
GEBV validation test problems (ssGBLUP)

- In our single-step test day model development: Interbull GEBV validation test results not satisfying (see e.g. Koivula et al. 2016 EAAP, Belfast)
  - Too low $b_1$ (and $R^2$)
  - Also the parent average validation is poor

Is this a problem of model or test?

The aim in this presentation is to take a better look on accuracy and stability of our evaluations (normal and genomic)
Nordic RDC Data

- Protein TD observations were used to derive 305d yields
  - single trait repeatability model for protein only
    \[ h^2 = 0.37 \] and \[ r = 0.55 \]

1. Animal model EBVs
2. Genomic animal model: single-step GEBVs.

The ss-GBLUP calculated
- with standard \( G_w \)-matrix, \( w = 0.10 \)
- full QT-transformation \( \) (Matilainen et al. 2016, Interbull, Chile)
- always applied for all the genotypes existing in whole data.

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Nordic RDC
Data reduced data sets

- Full data Data0
  - Calvings up to March 2016
  - 3.5M cows with 7M observations
  - Pedigree 5.4M animals, 33,321 genotyped

- Reduced data sets:
Model validation statistics

Interbull GEBV validation test

Reliability

Correlation(EBV1,EBV2) and regression EBV2 on EBV1 (SireType == Proven sires)

\[(G)EBV \text{ Stability} = \frac{(EBV2-EBV1)}{C-year} + Parity + SireType + \frac{(EBV2-EBV1)}{Parity \times SireType}\]

EBV1 is always the evaluation with less information, and EBV2 the following, next evaluation, with +1 record

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### Siretypes of cows by first year of calving (% of all used sires)

<table>
<thead>
<tr>
<th>Sire type</th>
<th>2010</th>
<th>2011</th>
<th>2012</th>
<th>2013</th>
<th>2014</th>
</tr>
</thead>
<tbody>
<tr>
<td>No daughters</td>
<td>30</td>
<td>37</td>
<td>44</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Progeny-tested</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>- Inseminations</td>
<td>61</td>
<td>54</td>
<td>49</td>
<td>42</td>
<td>16</td>
</tr>
</tbody>
</table>

**Distribution of sire age classes for cows born 2008, 2011, and 2014**

![Graph showing distribution of sire age classes](image-url)
GEBV validation test results
bulls, n=462

Regression of DYD\textsubscript{data0} on PA\textsubscript{data-3} or GEBV\textsubscript{data-3}

<table>
<thead>
<tr>
<th></th>
<th>PA</th>
<th>GEBV</th>
</tr>
</thead>
<tbody>
<tr>
<td>Protein</td>
<td>b\textsubscript{1} 0.75</td>
<td>b\textsubscript{1} 0.57</td>
</tr>
<tr>
<td></td>
<td>R\textsuperscript{2} 0.20</td>
<td>R\textsuperscript{2} 0.28</td>
</tr>
</tbody>
</table>

\[
R_{\text{validation}}^2 = \frac{R_{\text{model}}^2}{r_{\text{DYD}}^2}
\]
GEBV validation test results

Regression of YD to GEBV or EBV (PA)

<table>
<thead>
<tr>
<th>ProdYear</th>
<th>PA</th>
<th>GEBV</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>$b_1$</td>
<td>$R^2$</td>
</tr>
<tr>
<td>2012</td>
<td>0.95</td>
<td>0.22</td>
</tr>
<tr>
<td>2013</td>
<td>1.07</td>
<td>0.43</td>
</tr>
<tr>
<td>2014</td>
<td>1.01</td>
<td>0.38</td>
</tr>
</tbody>
</table>

$$R_{validation}^2 = \frac{R_{model}^2}{r_{YD}^2}$$

Note: Reference population in 2014 is much larger than in 2012.
Correlation between parent average (PA) and first lactation EBV

### Correlation of PA to 1. lactation EBV

<table>
<thead>
<tr>
<th>Prod year</th>
<th>EBV</th>
<th>mean</th>
<th>std</th>
<th>$r_{PA,1.Lact}$</th>
</tr>
</thead>
<tbody>
<tr>
<td>2012</td>
<td>PA</td>
<td>127</td>
<td>89</td>
<td>0.78</td>
</tr>
<tr>
<td></td>
<td>1.Lact EBV</td>
<td>130</td>
<td>109</td>
<td></td>
</tr>
<tr>
<td>2013</td>
<td>PA</td>
<td>130</td>
<td>92</td>
<td>0.79</td>
</tr>
<tr>
<td></td>
<td>1.Lact EBV</td>
<td>135</td>
<td>112</td>
<td></td>
</tr>
<tr>
<td>2014</td>
<td>PA</td>
<td>150</td>
<td>97</td>
<td>0.81</td>
</tr>
<tr>
<td></td>
<td>1.Lact EBV</td>
<td>156</td>
<td>120</td>
<td></td>
</tr>
</tbody>
</table>

### PA versus 1. lactation EBV by evaluation year of the PA

- **2012**: Slope = 0.94
- **2013**: Slope = 0.95
- **2014**: Slope = 0.97

Correlation for cows with SireType == "progeny-tested sire"
Let EBV1 include all information (~ERC1) in EBV2

Then

$$R_{EBV2}^2 = \frac{ERC1 + ERC2}{ERC1 + ERC2 + \lambda}$$

where $\lambda$ is the variance ratio

Because

$$r_{EBV1,EBV2}^2 = \frac{R_{EBV1}^2}{R_{EBV2}^2}$$


If you assume constant ERC2, e.g. ~ 0.9,

it is possible to solve ERC1 and thereafter $R_{EBV1}^2$
Correlation between parent average (PA) and first lactation EBV

**Correlation of PA to 1. lactation EBV**

<table>
<thead>
<tr>
<th>Prod year</th>
<th>EBV</th>
<th>mean</th>
<th>std</th>
<th>r_{PA,1}</th>
<th>R^2_{PA}</th>
</tr>
</thead>
<tbody>
<tr>
<td>2012</td>
<td>PA</td>
<td>127</td>
<td>89</td>
<td>0.78</td>
<td>0.31</td>
</tr>
<tr>
<td></td>
<td>1.Lact EBV</td>
<td>130</td>
<td>109</td>
<td>0.79</td>
<td>0.32</td>
</tr>
<tr>
<td>2013</td>
<td>PA</td>
<td>130</td>
<td>92</td>
<td>0.79</td>
<td>0.32</td>
</tr>
<tr>
<td></td>
<td>1.Lact EBV</td>
<td>135</td>
<td>112</td>
<td>0.81</td>
<td>0.34</td>
</tr>
<tr>
<td>2014</td>
<td>PA</td>
<td>150</td>
<td>97</td>
<td>0.81</td>
<td>0.34</td>
</tr>
<tr>
<td></td>
<td>1.Lact EBV</td>
<td>156</td>
<td>120</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Theoretically sire having 150 daughters and dam Nrec=2, \( R^2_{PA} \approx 0.37 \)

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Correlation between 1. lactation EBV and 2. lactation EBV within each production year

<table>
<thead>
<tr>
<th>Prodyear</th>
<th>EBV</th>
<th>mean</th>
<th>std</th>
<th>Corr</th>
</tr>
</thead>
<tbody>
<tr>
<td>2012</td>
<td>EBV1</td>
<td>122</td>
<td>104</td>
<td>0.93</td>
</tr>
<tr>
<td></td>
<td>EBV2</td>
<td>109</td>
<td>104</td>
<td></td>
</tr>
<tr>
<td>2013</td>
<td>EBV1</td>
<td>133</td>
<td>106</td>
<td>0.93</td>
</tr>
<tr>
<td></td>
<td>EBV2</td>
<td>128</td>
<td>107</td>
<td></td>
</tr>
<tr>
<td>2014</td>
<td>EBV1</td>
<td>140</td>
<td>109</td>
<td>0.94</td>
</tr>
<tr>
<td></td>
<td>EBV2</td>
<td>134</td>
<td>110</td>
<td></td>
</tr>
</tbody>
</table>

Correlation for cows with SireType == "progeny-tested sire"
LS means for EBV difference by lactation, siretype and production year (± 95%CL)

EBV difference = EBV2 – EBV1, change from evaluation to another

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Correlation with GEBV0 and first lactation GEBV

Correlation of GEBV0 to 1. lactation

<table>
<thead>
<tr>
<th>Prodyear</th>
<th>GEBV</th>
<th>mean</th>
<th>std</th>
<th>(r_{PA,1\text{.Lact}})</th>
<th>(R^{2}_{PA})</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Non-genotyped cows</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>2012</td>
<td>GEBV0</td>
<td>100</td>
<td>88.83</td>
<td>0.78</td>
<td>0.31</td>
</tr>
<tr>
<td></td>
<td>1.Lact</td>
<td>104</td>
<td>108</td>
<td></td>
<td></td>
</tr>
<tr>
<td>2013</td>
<td>GEBV0</td>
<td>105</td>
<td>85</td>
<td>0.72</td>
<td>0.24</td>
</tr>
<tr>
<td></td>
<td>1.Lact</td>
<td>114</td>
<td>110</td>
<td></td>
<td></td>
</tr>
<tr>
<td>2014</td>
<td>GEBV0</td>
<td>122</td>
<td>96</td>
<td>0.81</td>
<td>0.34</td>
</tr>
<tr>
<td></td>
<td>1.Lact</td>
<td>129</td>
<td>112</td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Genotyped cows</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>2012</td>
<td>GEBV0</td>
<td>157</td>
<td>127</td>
<td>0.93</td>
<td>0.56</td>
</tr>
<tr>
<td></td>
<td>1.Lact</td>
<td>153</td>
<td>126</td>
<td></td>
<td></td>
</tr>
<tr>
<td>2013</td>
<td>GEBV0</td>
<td>172</td>
<td>116</td>
<td>0.90</td>
<td>0.49</td>
</tr>
<tr>
<td></td>
<td>1.Lact</td>
<td>176</td>
<td>124</td>
<td></td>
<td></td>
</tr>
<tr>
<td>2014</td>
<td>GEBV0</td>
<td>192</td>
<td>122</td>
<td>0.93</td>
<td>0.56</td>
</tr>
<tr>
<td></td>
<td>1.Lact</td>
<td>189</td>
<td>129</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

GEBV-0 versus 1. lactation GEBV by evaluation year of the GEBV-0
LS means for difference GEBV2-GEBV1 by lactation, siretype and production year (± 95% CL)

GEBV difference = GEBV2 – GEBV1, change from evaluation to another
LS means for difference EBV2-EBV1 by lactation, siretype and production year (± 95%CL)

EBV difference = EBV2 – EBV1, change from evaluation to another

Siretype 1 = no daughters, Siretype 2 = progeny tested bull

Production year

LS means for EBV difference
Conclusions - use (G)EBV stability validation

Changes during consecutive evaluations with one year added can illustrate the stability of the evaluations
  - Especially useful when trait measured yearly at each animal

Use of females as validation group
  - Instead of one test (bulls) you can do yearly tests
  - Straight copy of current GEBV validation test has a weakness that the environmental effects (used to attain the DYD) are from the EBV model

Correlations between consecutive (G)EBVs reflected the accuracy
  - Correlation estimates consistent with expectations based on $R^2$
  - High correlation does not automatically mean high accuracy
Conclusions – RDC 305 d test evaluations

With simple 305 d animal model:
- Validation results for EBVs were at the low side
- Validation results for ssGBLUP GEBVs were also low
- Genomic selection might have affected accuracy of DYD?

Stability measured as year-to-year change was satisfactory
- Except for PA EBV, which increased --> EBV1 at the best 0.2 SD
- GEBV from ssGBLUP was much more stable
- General trend:
  • (G)EBV increases with 1\textsuperscript{st} lactation record,
  • (G)EBV decreases with 2\textsuperscript{nd} and 3\textsuperscript{rd} lactation records,
- True effect of GS not yet visible: selection 2010 not strong
Thank you!