National Screening Report

Germany 2012

Deutsche Gesellschaft für Neugeborenenscreening e.V.

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Abbreviations:
CAH Congenital adrenal hyperplasia
CACT - Deficiency Carnitin-Acylcarnitin-Translocase-Deficiency
CPT I - Deficiency Carnitin- Palmitoyl-CoA-Transferase I-Deficiency
CPT II - Deficiency Carnitin- Palmitoyl-CoA-Transferase II-Deficiency
GA I Glutaric acidaemia Type I
<table>
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</tr>
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</tr>
<tr>
<td>IVA</td>
<td>Isovaleric acidaemia</td>
</tr>
<tr>
<td>LCHAD - Deficiency</td>
<td>Long-Chain-3-Hydroxy-Acyl-CoA-Dehydrogenase-Deficiency</td>
</tr>
<tr>
<td>DoL</td>
<td>Day of life</td>
</tr>
<tr>
<td>DB</td>
<td>Dried blood</td>
</tr>
<tr>
<td>GV 1- 3</td>
<td>Guide value 1 - 3</td>
</tr>
<tr>
<td>MCAD - Deficiency</td>
<td>Medium-Chain-Acyl-CoA-Dehydrogenase-Deficiency</td>
</tr>
<tr>
<td>MSUD</td>
<td>Maple syrup urine disease</td>
</tr>
<tr>
<td>NGS</td>
<td>Newborn screening</td>
</tr>
<tr>
<td>SV</td>
<td>Secondary value</td>
</tr>
<tr>
<td>PKU</td>
<td>Phenylketonuria</td>
</tr>
<tr>
<td>PPV</td>
<td>Positive predictive value</td>
</tr>
<tr>
<td>Second-tier Process</td>
<td>In suspicious results secondary analysis of additional parameter or alternative analytical methods from the same test cards</td>
</tr>
<tr>
<td>WoG</td>
<td>Week of gestation</td>
</tr>
<tr>
<td>VLCAD - Deficiency</td>
<td>Very-Long-Chain-Acyl-CoA-Dehydrogenase-Deficiency</td>
</tr>
</tbody>
</table>
Screening Laboratories and Screening Centres

Screening Centres (laboratories) with different localities or laboratories which are connected to a screening centre are analysed stratified.

(1) Neugeborenen Screeninglabor Berlin
Dr. med. Oliver Blankenstein
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sabine.roenicke@med.ovgu.de
http://www.stoffwechselzentrum-magdeburg.de

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rgesundheit/neugeborenenscreening/index.htm
1 Introduction

The newborn screening is a medical population based preventative measure with the aim of early and sufficient detection and high quality therapy of all newborns with treatable endocrine metabolic diseases.

The guidelines of prevention of disease for children up to 6 years of age („Kinder-Richtlinien“) outline the details of newborn screening (NBS) in the appendices 2-4.

The National Screening Report was composed by the “Deutschen Gesellschaft für Neugeborenscreening (DGNS e.V.)” as well as the German screening laboratories. The statistical analysis of the screening data was according to the guidelines and their quality criteria of the NBS implementation. This report targets only the metabolic and endocrine diseases which are defined in these guidelines. It provides a wide statistical summary of disease related screening numbers and recall numbers at diagnoses for the year 2012. Additionally, data for process quality are presented.

Process quality describes the process flow and its evaluation through specialists according to defined indicators. These are the following for the newborn screening:

- Total survey of the targeted population
  - Collection method and rate
  - Blank card system
- Completeness of the control and the follow-up studies
- Collection of test parameters and cut offs
- Stratified rates of recall, positive predictive values and prevalence according to laboratory, age as well as gestational age,
- Specificity and sensitivity of diagnostic tests
- Process times (pre analytic and laboratory), age at blood collection, time within blood collections, time of arrival in the laboratory and time of result communication
- Screening values of newborns for which further testing is emphasized
- Diagnostics for confirmation
  - Type of diagnostics
  - Time of diagnostics
- Final diagnosis
- Start of therapy

Previously, laboratories were listed which have undertaken the screening in 2012 for Germany. (12 and 13 relate to the same laboratory, one with and without the cooperation of the Screening Centre, same for 14 and 15). In the tables the laboratories are encrypted. Paragraphs in the text relate to the guidelines for children from 16.12.2010 [1]. Tables are numbered according to the chapters.

We would like to thank all the laboratories for provision of their data. The data was checked for plausibility. Remaining inconsistencies of data was analysed according to the reported data. (Inconsistencies can sometimes be due to the system).
The screening samples of the federal states are spread to the laboratories according to Figure 1.

**Figure 1: Distribution of analysis according to county and laboratory**
2 Results

In the year 2012, 673.544 children were born in Germany [2]. The total recorded screening of 674.926 exceeds this number. A cause for the additional screening cards cannot be declared. Reasons could be, not as such, registered repeat screening cards or cards of births not registered in Germany. Further investigations cannot be undertaken as data exchange is not legalised.

Births [2]: 673.544
First screening: 674.926
Final diagnosis (see Table 3): 516

A secure statement about the rate of participation in NBS can only be made by comparison of person related data or the population. By law this is only legal in the county of Bavaria.

In the German guidelines, the targeted diseases are defined for the nationwide screening. Some laboratories will also screen for scientific purposes. These results will not be addressed in this report. Out of 1 in 1.305 newborns, one targeted disease according to the guidelines is found. Table 2 shows the prevalence of targeted diseases in the year 2012 in Germany.

<table>
<thead>
<tr>
<th>Disease</th>
<th>Confirmed cases</th>
<th>Prevalence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hypothyroidism</td>
<td>205</td>
<td>1: 3 285</td>
</tr>
<tr>
<td>Congenital adrenal hyperplasia (CAH)</td>
<td>48</td>
<td>1: 14 032</td>
</tr>
<tr>
<td>Biotinidase deficiency (incl. partial defect)</td>
<td>37</td>
<td>1: 18 204</td>
</tr>
<tr>
<td>Galactosaemia (classic)</td>
<td>9</td>
<td>1: 74 838</td>
</tr>
<tr>
<td>Phenylketonuria (PKU) n=63 / Hyperphenylalaninaemia(HPA) n=63</td>
<td>126</td>
<td>1: 5 388</td>
</tr>
<tr>
<td>Maple syrup urine disease (MSUD)</td>
<td>5</td>
<td>1: 134 709</td>
</tr>
<tr>
<td>Medium-Chain-Acyl-CoA-Dehydrogenase (MCAD)-Deficiency</td>
<td>62</td>
<td>1: 10 864</td>
</tr>
<tr>
<td>Long-Chain-3-OH-Acyl-CoA-Dehydrogenase (LCHAD)-Deficiency</td>
<td>7</td>
<td>1: 96 221</td>
</tr>
<tr>
<td>(Very-)Long-Chain-Acyl-CoA-Dehydrogenase (VLCAD)-Deficiency</td>
<td>7</td>
<td>1: 96 221</td>
</tr>
<tr>
<td>Carnitin-Palmitoyl-CoA-Transferase I (CPT I)-Deficiency</td>
<td>0</td>
<td></td>
</tr>
<tr>
<td>Carnitin-Palmitoyl-CoA-Transferase II (CPT II)-Deficiency</td>
<td>0</td>
<td></td>
</tr>
<tr>
<td>Carnitin-Acylcarnitin-Translocase (CACT)-Deficiency</td>
<td>0</td>
<td></td>
</tr>
<tr>
<td>Glutaric aciduria Type I (GA I)</td>
<td>5</td>
<td>1: 134 709</td>
</tr>
<tr>
<td>Isovalerianacidaemia (IVA)</td>
<td>5</td>
<td>1: 134 709</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>516</strong></td>
<td>1: 1 307</td>
</tr>
</tbody>
</table>
2.1 Data of primary screening

According to the guidelines of children, every newborn should be screened before leaving the birth facility. A reliable screening can only be undertaken with blood sampling beyond the completed 32nd gestational week and 36th hour of life. A primary screening before the 36th hour of life or before the completed 32nd week of gestation should be followed by a repeat screening. The following table shows the stratified results of the primary screening according to age and gestational age.

Table 2.1: Age at primary screening

<table>
<thead>
<tr>
<th>Lab</th>
<th>Total</th>
<th>n</th>
<th>%</th>
<th>n</th>
<th>%</th>
<th>n</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>≥36h and ≥32WoG</td>
<td></td>
<td>&lt;36h and ≥32WoG</td>
<td></td>
<td>&lt;32WoG</td>
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<tr>
<td>1</td>
<td>52534</td>
<td>50690</td>
<td>96.49</td>
<td>1152</td>
<td>2.19</td>
<td>692</td>
<td>1.32</td>
</tr>
<tr>
<td>3</td>
<td>15538</td>
<td>15229</td>
<td>98.01</td>
<td>160</td>
<td>1.03</td>
<td>149</td>
<td>0.96</td>
</tr>
<tr>
<td>5</td>
<td>52547</td>
<td>51137</td>
<td>97.32</td>
<td>847</td>
<td>1.61</td>
<td>563</td>
<td>1.07</td>
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<tr>
<td>6</td>
<td>12742</td>
<td>12275</td>
<td>96.33</td>
<td>315</td>
<td>2.47</td>
<td>152</td>
<td>1.19</td>
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<tr>
<td>7</td>
<td>45018</td>
<td>43525</td>
<td>96.68</td>
<td>774</td>
<td>1.72</td>
<td>719</td>
<td>1.60</td>
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<tr>
<td>8</td>
<td>156526</td>
<td>153215</td>
<td>97.88</td>
<td>1534</td>
<td>0.98</td>
<td>1777</td>
<td>1.14</td>
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<tr>
<td>9</td>
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<td>107914</td>
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<td>1591</td>
<td>1.44</td>
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<td>10</td>
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<td>296</td>
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<td>12a</td>
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<td>13</td>
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<td>658107</td>
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<td>8686</td>
<td>1.29</td>
<td>8133</td>
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</tbody>
</table>

* Including screening tests that have not been sent to the tracking center (without consent)

2.2 Relation of requested to received repeat screenings

In Table 2.2.1 the repeat screenings are listed stratified according to their base of request defined as:

- **"<32WoG"**: all sample of newborns before 32 WoG, independent of age and result of primary screening
- **"<36h"**: all sample of newborns beyond 32 WoG, but age less than 36h, independent of the result of primary screening
- **Recall**: essential repeat testing due to suspicious primary screening at a gestational age > 32 WoG and age > 36h
### Table 2.2: Requested and received repeat screenings

<table>
<thead>
<tr>
<th>Lab</th>
<th>Total&lt;sup&gt;a&lt;/sup&gt;&lt;sup&gt;c&lt;/sup&gt; requested</th>
<th>Total&lt;sup&gt;a&lt;/sup&gt; received</th>
<th>%</th>
<th>Recall requested&lt;sup&gt;c&lt;/sup&gt;</th>
<th>Recall received</th>
<th>%</th>
</tr>
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<tr>
<td>1</td>
<td>2436</td>
<td>2356</td>
<td>96.72</td>
<td>645</td>
<td>632</td>
<td>97.98</td>
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<tr>
<td>3&lt;sup&gt;b&lt;/sup&gt;</td>
<td>400</td>
<td>470</td>
<td>117.50&lt;sup&gt;d&lt;/sup&gt;</td>
<td>98</td>
<td>160</td>
<td>163.27&lt;sup&gt;d&lt;/sup&gt;</td>
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<tr>
<td>5</td>
<td>1865</td>
<td>1818</td>
<td>97.48</td>
<td>455</td>
<td>418</td>
<td>91.87</td>
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<tr>
<td>6</td>
<td>548</td>
<td>548</td>
<td>100.00</td>
<td>62</td>
<td>62</td>
<td>100.00</td>
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<tr>
<td>7&lt;sup&gt;b&lt;/sup&gt;</td>
<td>2352</td>
<td>n.s.</td>
<td>n.s.</td>
<td>875</td>
<td>n.s.</td>
<td>n.s.</td>
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<td>100.00</td>
</tr>
<tr>
<td>15</td>
<td>427</td>
<td>326</td>
<td>76.35</td>
<td>84</td>
<td>81</td>
<td>96.43</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>22225</strong></td>
<td><strong>18296</strong></td>
<td><strong>93.96</strong>&lt;sup&gt;b&lt;/sup&gt;</td>
<td><strong>5082</strong></td>
<td><strong>4045</strong></td>
<td><strong>98.44</strong>&lt;sup&gt;b&lt;/sup&gt;</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Lab</th>
<th>&lt;36h requested&lt;sup&gt;c&lt;/sup&gt;</th>
<th>&lt;36h received</th>
<th>%</th>
<th>&lt;32WOG requested&lt;sup&gt;c&lt;/sup&gt;</th>
<th>&lt;32WOG received</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>1033</td>
<td>1033</td>
<td>100.00</td>
<td>673</td>
<td>608</td>
<td>90.34</td>
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<td>3&lt;sup&gt;b&lt;/sup&gt;</td>
<td>160</td>
<td>170</td>
<td>106.25&lt;sup&gt;d&lt;/sup&gt;</td>
<td>138</td>
<td>140</td>
<td>101.45&lt;sup&gt;d&lt;/sup&gt;</td>
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<td>563</td>
<td>556</td>
<td>98.76</td>
</tr>
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<td>314</td>
<td>314</td>
<td>100.00</td>
<td>146</td>
<td>146</td>
<td>100.00</td>
</tr>
<tr>
<td>7&lt;sup&gt;b&lt;/sup&gt;</td>
<td>771</td>
<td>n.s.</td>
<td>n.s.</td>
<td>706</td>
<td>n.s.</td>
<td>n.s.</td>
</tr>
<tr>
<td>8</td>
<td>1525</td>
<td>1348</td>
<td>88.39</td>
<td>1725</td>
<td>1604</td>
<td>92.99</td>
</tr>
<tr>
<td>9</td>
<td>1154</td>
<td>728</td>
<td>63.08</td>
<td>1586</td>
<td>1224</td>
<td>77.18</td>
</tr>
<tr>
<td>10</td>
<td>348</td>
<td>343</td>
<td>98.56</td>
<td>280</td>
<td>280</td>
<td>100.00</td>
</tr>
<tr>
<td>11</td>
<td>361</td>
<td>353</td>
<td>97.78</td>
<td>155</td>
<td>155</td>
<td>100.00</td>
</tr>
<tr>
<td>12</td>
<td>873</td>
<td>865</td>
<td>99.08</td>
<td>806</td>
<td>806</td>
<td>100.00</td>
</tr>
<tr>
<td>13</td>
<td>808</td>
<td>787</td>
<td>97.40</td>
<td>824</td>
<td>824</td>
<td>100.00</td>
</tr>
<tr>
<td>14</td>
<td>219</td>
<td>218</td>
<td>99.54</td>
<td>184</td>
<td>184</td>
<td>100.00</td>
</tr>
<tr>
<td>15</td>
<td>135</td>
<td>53</td>
<td>39.26</td>
<td>173</td>
<td>157</td>
<td>90.75</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td><strong>8548</strong></td>
<td><strong>7056</strong></td>
<td><strong>92.63</strong>&lt;sup&gt;b&lt;/sup&gt;</td>
<td><strong>7959</strong></td>
<td><strong>6684</strong></td>
<td><strong>93.94</strong>&lt;sup&gt;b&lt;/sup&gt;</td>
</tr>
</tbody>
</table>

<sup>a</sup> Including secondary screening due to blood transfusion, parenteral nutrition or medication
<sup>b</sup> Calculation without labs giving not differentiated numbers
<sup>c</sup> Deaths are not included in the number of requested samples
<sup>d</sup> Figures have been confirmed by the lab
2.3 Tracking of completeness of screening

The newborn screening is a measure of public health and should be given to all German born children. To guarantee that the screening is offered to all newborns the tracking of completeness is necessary. For children born in obstetric units, control can be undertaken through hospital records or if permitted by state law through the birth registry.

Currently both measures are not undertaken nationwide. To target the tracking of completeness the following rule was included into the “guidelines”. The obstetric unit should document on a blank test card refusal of screening or death of a neonate. This test card should then be sent to the screening centre. The laboratories receive blank test cards in various numbers. The number of blank screening cards due to refusal is still low but higher than the previous years and in 2012 partly doubled compared to 2010. To what extent this is due to, a better reply or raised true refusals, is not clear.

This system seems to work mainly with the refusals respectively the declined early screening. Both, before screening deceased and the transferred neonates, would give expectations to higher numbers.

<table>
<thead>
<tr>
<th>Lab</th>
<th>Primary screening Total</th>
<th>Reasons for blank cards</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>Deceased</td>
</tr>
<tr>
<td></td>
<td>n</td>
<td>n</td>
</tr>
<tr>
<td>1</td>
<td>52534</td>
<td>48</td>
</tr>
<tr>
<td>3</td>
<td>15538</td>
<td>22</td>
</tr>
<tr>
<td>5</td>
<td>52547</td>
<td>82</td>
</tr>
<tr>
<td>6</td>
<td>12742</td>
<td>22</td>
</tr>
<tr>
<td>7*</td>
<td>45018</td>
<td>n.s.</td>
</tr>
<tr>
<td>8</td>
<td>156526</td>
<td>n.s.</td>
</tr>
<tr>
<td>9*</td>
<td>110661</td>
<td>8</td>
</tr>
<tr>
<td>10</td>
<td>34621</td>
<td>166</td>
</tr>
<tr>
<td>11</td>
<td>16666</td>
<td>51</td>
</tr>
<tr>
<td>12</td>
<td>80367</td>
<td>8</td>
</tr>
<tr>
<td>13*</td>
<td>64346</td>
<td>n.s.</td>
</tr>
<tr>
<td>14</td>
<td>25207</td>
<td>0</td>
</tr>
<tr>
<td>15*</td>
<td>8153</td>
<td>n.s.</td>
</tr>
<tr>
<td>Total</td>
<td>674926</td>
<td>407</td>
</tr>
</tbody>
</table>

\* Including early screening, transferral, no reason (declared)

\* No tracking of blank screening cards

\* No reason declared
Table 2.4: Secondary screening card due to poor sample quality

<table>
<thead>
<tr>
<th>Lab</th>
<th>Primary screening</th>
<th>Control requested</th>
<th>Control received</th>
<th>received/ requested (%)</th>
<th>Proportion of Primary screening (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>52534</td>
<td>418</td>
<td>392</td>
<td>93.78</td>
<td>0.80</td>
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<td>3</td>
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<td>46</td>
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<td>5</td>
<td>52547</td>
<td>466</td>
<td>460</td>
<td>98.71</td>
<td>0.89</td>
</tr>
<tr>
<td>6</td>
<td>12742</td>
<td>15</td>
<td>15</td>
<td>100.0</td>
<td>0.12</td>
</tr>
<tr>
<td>7</td>
<td>45018</td>
<td>n.s.</td>
<td>n.s.</td>
<td>n.s.</td>
<td>n.s.</td>
</tr>
<tr>
<td>8</td>
<td>156526</td>
<td>233</td>
<td>228</td>
<td>97.85</td>
<td>0.15</td>
</tr>
<tr>
<td>9</td>
<td>110661</td>
<td>547</td>
<td>488</td>
<td>89.21</td>
<td>0.49</td>
</tr>
<tr>
<td>10</td>
<td>34621</td>
<td>105</td>
<td>101</td>
<td>96.19</td>
<td>0.30</td>
</tr>
<tr>
<td>11</td>
<td>16666</td>
<td>2</td>
<td>2</td>
<td>100.00</td>
<td>0.01</td>
</tr>
<tr>
<td>12</td>
<td>80367</td>
<td>290</td>
<td>283</td>
<td>97.59</td>
<td>0.36</td>
</tr>
<tr>
<td>13</td>
<td>64346</td>
<td>245</td>
<td>243</td>
<td>99.18</td>
<td>0.38</td>
</tr>
<tr>
<td>14</td>
<td>25207</td>
<td>24</td>
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<td>15</td>
<td>8153</td>
<td>4</td>
<td>4</td>
<td>100.00</td>
<td>0.05</td>
</tr>
<tr>
<td>Total</td>
<td>674926</td>
<td>2395</td>
<td>2286</td>
<td>95.45</td>
<td>0.38*</td>
</tr>
</tbody>
</table>

* Calculation without Lab 7, since no mention of cards with poor sample quality

3 Quality of the screening analysis

The excellence of a test is measured by the sensitivity, the specificity as well as the positive predictive value. In screening, the sensitivity (true-test positives) but more so the specificity (true-test negatives), should be high to find all diseases and to avoid unnecessary worries and costs. The lower the rate of necessary control screening due to positive first screening (recall rate) the higher the specificity. In 2012 the recall rate was 0.78%. If we consider only screening cards of term newborns sampled beyond the 36th hour of life, the recall rate is 0.58%, meaning of 1000 tests only 6 are recalled. With sampling before the 36th hour of life or the 32nd WoG a secondary screening has to be done irrespectively of the results.

The total specificity was 99.29%. The sensitivity cannot be quoted, because systematic registration of unscreened neonates is not done.
Table 3: Recall rates and cases found for Germany 2012 N= 674,926*

<table>
<thead>
<tr>
<th>Disease</th>
<th>Recall ≥36h n</th>
<th>(%)</th>
<th>Recall &lt;36h n</th>
<th>(%)</th>
<th>Recall &lt;32WOG n</th>
<th>(%)</th>
<th>Recall Total n</th>
<th>(%)</th>
<th>Not found in the screening n</th>
<th>(%)</th>
<th>Confirmed cases n</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hypothyroidism</td>
<td>457</td>
<td>0.07</td>
<td>348</td>
<td>4.01</td>
<td>48</td>
<td>0.59</td>
<td>5</td>
<td>0.13</td>
<td>205</td>
<td></td>
<td></td>
</tr>
<tr>
<td>CAH</td>
<td>2098</td>
<td>0.32</td>
<td>360</td>
<td>4.14</td>
<td>541</td>
<td>6.65</td>
<td>1</td>
<td>0.44</td>
<td>48</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Biotinidase-Deficiency</td>
<td>156</td>
<td>0.02</td>
<td>9</td>
<td>0.10</td>
<td>20</td>
<td>0.25</td>
<td>0</td>
<td>0.03</td>
<td>37</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Classic Galactosaemia</td>
<td>221</td>
<td>0.03</td>
<td>5</td>
<td>0.06</td>
<td>8</td>
<td>0.10</td>
<td>0</td>
<td>0.03</td>
<td>9</td>
<td></td>
<td></td>
</tr>
<tr>
<td>PKU/HPA</td>
<td>210</td>
<td>0.03</td>
<td>16</td>
<td>0.18</td>
<td>33</td>
<td>0.41</td>
<td>0</td>
<td>0.04</td>
<td>126</td>
<td></td>
<td></td>
</tr>
<tr>
<td>MSUD</td>
<td>68</td>
<td>0.01</td>
<td>1</td>
<td>0.01</td>
<td>5</td>
<td>0.06</td>
<td>0</td>
<td>0.01</td>
<td>5</td>
<td></td>
<td></td>
</tr>
<tr>
<td>MCAD</td>
<td>130</td>
<td>0.02</td>
<td>4</td>
<td>0.05</td>
<td>3</td>
<td>0.04</td>
<td>0</td>
<td>0.02</td>
<td>62</td>
<td></td>
<td></td>
</tr>
<tr>
<td>LCHAD</td>
<td>53</td>
<td>0.01</td>
<td>0</td>
<td>0</td>
<td>2</td>
<td>0.02</td>
<td>0</td>
<td>0.01</td>
<td>7</td>
<td></td>
<td></td>
</tr>
<tr>
<td>VLCAD</td>
<td>173</td>
<td>0.03</td>
<td>3</td>
<td>0.03</td>
<td>6</td>
<td>0.07</td>
<td>0</td>
<td>0.03</td>
<td>7</td>
<td></td>
<td></td>
</tr>
<tr>
<td>CPT I-Deficiency</td>
<td>10</td>
<td>0.0015</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0.0015</td>
<td>0</td>
<td>0.0015</td>
<td>0</td>
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<td>0</td>
</tr>
<tr>
<td>CPT II-Deficiency</td>
<td>33</td>
<td>0.01</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0.01</td>
<td>0</td>
<td>0.01</td>
<td>0</td>
<td></td>
<td></td>
</tr>
<tr>
<td>CAT-Deficiency</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
<td></td>
<td></td>
</tr>
<tr>
<td>GA I</td>
<td>180</td>
<td>0.03</td>
<td>2</td>
<td>0.02</td>
<td>10</td>
<td>0.12</td>
<td>0</td>
<td>0.03</td>
<td>5</td>
<td></td>
<td></td>
</tr>
<tr>
<td>IVA</td>
<td>53</td>
<td>0.01</td>
<td>0</td>
<td>0</td>
<td>9</td>
<td>0.11</td>
<td>0</td>
<td>0.01</td>
<td>5</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Total</td>
<td>3842</td>
<td>0.58</td>
<td>748</td>
<td>8.61</td>
<td>685</td>
<td>8.42</td>
<td>0.78</td>
<td>6</td>
<td>516</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

* Primary screening Total: n= 674,926; Primary screening ≥ 36h and ≥ 32WOG n=658,107; Primary screening <36h n=8,686; Primary screening < 32WOG n=8,133

a All preterm infants under 32 WOG
b Mutation: I172N

3.1 Recall rate and confirmed cases stratified

The following tables show recall rates ≥36h and confirmed cases stratified for the Laboratory. The reference of > 36 hours automatically includes > 32 weeks gestational age. The confirmed diagnosis, confirmed cases and their prevalence relate to the total screening tests, irrespectively of age and gestational age. The validation of confirmed cases was tested for plausibility of metabolic diseases by Professor Andreas Schulze and Dr. Regina Ensenauer, for endocrine diseases by Dr. Oliver Blankenstein and PD Dr. Heiko Krude. Excluded and therefore not reported are cases with missing data of confirmation diagnostics (n=21) (Tab.3.1.a) and cases where the confirmation diagnostics were negative (n=4). For some diseases the true prevalence could be higher. Double reported cases were included only once.
Table 3.1 : Cases with missing data of confirmation diagnostics

<table>
<thead>
<tr>
<th>Disease</th>
<th>Data missing</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hypothyroidism</td>
<td>11</td>
</tr>
<tr>
<td>CAH</td>
<td>3</td>
</tr>
<tr>
<td>Biotinidase deficiency</td>
<td>1</td>
</tr>
<tr>
<td>MCAD</td>
<td>3</td>
</tr>
<tr>
<td>VLCAD</td>
<td>2</td>
</tr>
<tr>
<td>GA I</td>
<td>1</td>
</tr>
<tr>
<td>Total</td>
<td>21</td>
</tr>
</tbody>
</table>

In the following tables Recall rates <0.01% and very small n are not calculated, small values of large differences would show influence

### 3.1.1 Hypothyroidism

<table>
<thead>
<tr>
<th>Lab</th>
<th>Primary screening Total</th>
<th>Primary screening ≥36h</th>
<th>Recall ≥36h</th>
<th>Recall-rate(%)</th>
<th>Confirmed cases*</th>
<th>Not found in the screening</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>52534</td>
<td>50690</td>
<td>32</td>
<td>0.06</td>
<td>18</td>
<td>1</td>
</tr>
<tr>
<td>3</td>
<td>15538</td>
<td>15229</td>
<td>10</td>
<td>0.07</td>
<td>7</td>
<td>0</td>
</tr>
<tr>
<td>5</td>
<td>52547</td>
<td>51137</td>
<td>52</td>
<td>0.10</td>
<td>21</td>
<td>1</td>
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<tr>
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<td>12742</td>
<td>12275</td>
<td>5</td>
<td>0.04</td>
<td>3</td>
<td>0</td>
</tr>
<tr>
<td>7</td>
<td>45018</td>
<td>43525</td>
<td>7</td>
<td>0.02</td>
<td>5</td>
<td>0</td>
</tr>
<tr>
<td>8</td>
<td>156526</td>
<td>153215</td>
<td>168</td>
<td>0.11</td>
<td>68</td>
<td>3</td>
</tr>
<tr>
<td>9</td>
<td>110661</td>
<td>107914</td>
<td>59</td>
<td>0.05</td>
<td>30</td>
<td>0</td>
</tr>
<tr>
<td>10</td>
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<td>33975</td>
<td>26</td>
<td>0.08</td>
<td>12</td>
<td>0</td>
</tr>
<tr>
<td>11</td>
<td>16666</td>
<td>16145</td>
<td>13</td>
<td>0.08</td>
<td>3</td>
<td>0</td>
</tr>
<tr>
<td>12</td>
<td>80367</td>
<td>78640</td>
<td>31</td>
<td>0.04</td>
<td>20</td>
<td>0</td>
</tr>
<tr>
<td>13</td>
<td>64346</td>
<td>62714</td>
<td>27</td>
<td>0.04</td>
<td>11</td>
<td>0</td>
</tr>
<tr>
<td>14</td>
<td>25207</td>
<td>24798</td>
<td>16</td>
<td>0.06</td>
<td>5</td>
<td>0</td>
</tr>
<tr>
<td>15</td>
<td>8153</td>
<td>7850</td>
<td>11</td>
<td>0.14</td>
<td>3</td>
<td>0</td>
</tr>
<tr>
<td>Total</td>
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<td>658107</td>
<td>457</td>
<td>0.07</td>
<td>205</td>
<td>5*</td>
</tr>
</tbody>
</table>

* Including transient hypothyroidism n =4,

a All premature babies born under 32 WoG (see Table 5.2)

In addition n=7 hyperthyrotropinaemia reported and confirmed. These are not included in the calculation of prevalence.
### 3.1.2 Congenital adrenal hyperplasia (CAH)

<table>
<thead>
<tr>
<th>Lab</th>
<th>Primary screening Total</th>
<th>Primary screening ≥36h</th>
<th>Recall ≥36h</th>
<th>Recall rate(%)</th>
<th>Confirmed cases</th>
<th>Not found in the screening</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>52534</td>
<td>50690</td>
<td>138</td>
<td>0.27</td>
<td>4</td>
<td>0</td>
</tr>
<tr>
<td>3</td>
<td>15538</td>
<td>15229</td>
<td>5</td>
<td>0.03</td>
<td>2</td>
<td>0</td>
</tr>
<tr>
<td>5</td>
<td>52547</td>
<td>51137</td>
<td>342</td>
<td>0.67</td>
<td>2</td>
<td>0</td>
</tr>
<tr>
<td>6</td>
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<td>12275</td>
<td>25</td>
<td>0.20</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>7</td>
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<td>0.98</td>
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<td>0</td>
</tr>
<tr>
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<td>153215</td>
<td>63</td>
<td>0.04</td>
<td>16</td>
<td>0</td>
</tr>
<tr>
<td>9</td>
<td>110661</td>
<td>107914</td>
<td>246</td>
<td>0.23</td>
<td>11</td>
<td>0</td>
</tr>
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<td>10</td>
<td>34621</td>
<td>33975</td>
<td>155</td>
<td>0.46</td>
<td>2</td>
<td>1&lt;sup&gt;b&lt;/sup&gt;</td>
</tr>
<tr>
<td>11</td>
<td>16666</td>
<td>16145</td>
<td>39</td>
<td>0.24</td>
<td>4</td>
<td>0</td>
</tr>
<tr>
<td>12</td>
<td>80367</td>
<td>78640</td>
<td>373</td>
<td>0.47</td>
<td>4</td>
<td>0</td>
</tr>
<tr>
<td>13</td>
<td>64346</td>
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<sup>a</sup> Laboratory used second-tier process  
<sup>b</sup> Genetics: I172N

### 3.1.3 Biotinidase deficiency

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<sup>*</sup> Recall rate recorded only if ≥ 0.01% and n > 5.
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* Recall rate recorded only if ≥ 0.01% and n > 5.
** Variants are not comprehensively covered

### 3.1.5 PKU / HPA

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* Recall rate recorded only if ≥ 0.01% and n > 5.
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* Recall rate recorded only if ≥ 0.01% and n > 5.

### 3.1.7 MCAD-Deficiency

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* Recall rate recorded only if ≥ 0.01% and n > 5.
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* Recall rate recorded only if ≥ 0.01% and n > 5.

### 3.1.9 VLCAD-Deficiency

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* Recall rate recorded only if ≥ 0.01% and n > 5.
### 3.1.10 No confirmed cases of CPTI-Deficiency and for CPT II-Deficiency

### 3.1.11 Glutaric aciduria Type I

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<th>Recall rate(%)*</th>
<th>Confirmed cases</th>
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* Recall rate recorded only if $\geq 0.01\%$ and $n > 5$.

### 3.1.12 Isovaleric acidaemia

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<th>Recall rate(%)*</th>
<th>Confirmed cases</th>
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* Recall rate recorded only if $\geq 0.01\%$ and $n > 5$. 
3.2 Recall rate stratified according to time of primary screening

The number of positives, especially false positive screening results and therefore the recall rate depends on age and gestational age. Earlier testing than the 36th hour of life and a gestational age of <32 weeks increases the risk of false negative and false positive results. This differs for the targeted diseases. In the following tables we stratify the recall rates by gestational age and timing of the sampling. Recall rate is recorded only if it exceeds 0.01% and n > 5 since small numbers cause a high variability.

3.2.1 Hypothyroidism

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<th>Primary screening &lt; 36h</th>
<th>Primary screening &lt; 32WoG</th>
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* Laboratory used second-tier process at screening >36h and <32 WoG

### 3.2.3 Biotinidase deficiency

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### 3.2.5 PKU/HPA

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### 3.2.7 MCAD-Deficiency

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DGNS-Report 2012  Page 25 of 43
### 3.2.8 LCHAD-Deficiency

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### 3.2.12 CACT-Deficiency

No recalls for the CACT deficiency were reported in 2012

### 3.2.13 Glutaric aciduria Type I

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4 Process Periods

4.1 Age at blood collection

According to the guidelines (§8.1) of children, every newborn should be screened beyond the completed 32nd gestational week and 36th hour of life. In 90% of cases, with specification of collection time, the collection was according to the guidelines, in 8.65% (3.15-12.03%) beyond the 72nd hour of life, in 11.56% (1.02-3.72%) before the 36th hour of life (see Table 4.1). The proportion of samples which were sampled after 72 hours could be lowered from 22.25 % in 2005 to 9.67 % in 2012 (see figure 2). These numbers clearly imply an improvement of the process quality, since the adherence to the optimal timeframe is of great importance to the efficiency of the screening. Life threatening metabolic or electrolyte crisis can be prevented by early diagnosis and therapy.

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<td>7738</td>
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<td>9734</td>
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<td>2043</td>
<td>8.40</td>
</tr>
<tr>
<td>15</td>
<td>8153</td>
<td>135</td>
<td>1.66</td>
<td>3737</td>
<td>45.84</td>
<td>3664</td>
<td>44.94</td>
<td>617</td>
<td>7.57</td>
</tr>
<tr>
<td>Total</td>
<td>661465</td>
<td>10315</td>
<td>1.56</td>
<td>247237</td>
<td>37.38</td>
<td>346668</td>
<td>52.41</td>
<td>57245</td>
<td>8.65</td>
</tr>
</tbody>
</table>

Due to missing data the stated number is smaller than the total number of primary screening. (marked with a).
4.2 Period from sampling to laboratory receipt

The time span between sampling and report of suspect results should not exceed 72 hours (paragraph 6. section 3). In 21.75% of cases with statement of the delivery time the probe was received 72 hours after sampling, in 23.55% of the cases between 48 and 72 hours. Shorter periods of delivery times are desirable, especially at the weekend. (Table 4.2)

Table 4.2: Period from sampling to laboratory receipt

<table>
<thead>
<tr>
<th>Lab</th>
<th>n</th>
<th>≤24h</th>
<th>%</th>
<th>&gt;24h-48h</th>
<th>%</th>
<th>&gt;48h-72h</th>
<th>%</th>
<th>&gt;72h</th>
<th>%</th>
</tr>
</thead>
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<td>14037</td>
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<td>37.86</td>
<td>10749</td>
<td>20.52</td>
<td>7773</td>
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<td>6155</td>
<td>39.61</td>
<td>5828</td>
<td>37.51</td>
<td>2362</td>
<td>15.20</td>
<td>1193</td>
<td>7.68</td>
</tr>
<tr>
<td>5</td>
<td>52552</td>
<td>6456</td>
<td>12.28</td>
<td>22630</td>
<td>43.06</td>
<td>14162</td>
<td>26.95</td>
<td>9304</td>
<td>17.70</td>
</tr>
<tr>
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<td>2062</td>
<td>17.34</td>
<td>5324</td>
<td>44.77</td>
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<td>25.78</td>
<td>1440</td>
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</tr>
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<td>30.88</td>
<td>8345</td>
<td>18.54</td>
<td>8490</td>
<td>18.86</td>
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<td>16951</td>
<td>11.32</td>
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<td>28.07</td>
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<td>26.27</td>
<td>40530</td>
<td>36.63</td>
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<td>36.75</td>
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<td>22.80</td>
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<td>35.15</td>
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<td>55.74</td>
<td>6767</td>
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<td>2141</td>
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<td>1752</td>
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<td>34.41</td>
<td>156518</td>
<td>23.55</td>
<td>144557</td>
<td>21.75</td>
</tr>
</tbody>
</table>

Due to missing data the stated number is smaller than the total number of primary screening of the previous tables (marked with *)
4.3 Period between laboratory receipt and result reporting

In 82% of probes the results get reported within 24 hours. The process time in borderline elevated results can be prolonged due to repeat testing (quality control) (Table 4.3 Figure 4).

Table 4.3 Period between laboratory receipt and result reporting

<table>
<thead>
<tr>
<th>Lab</th>
<th>Total</th>
<th>≤24h</th>
<th>%</th>
<th>&gt;24h-48h</th>
<th>&gt;48h-72h</th>
<th>&gt;72h</th>
<th>%</th>
</tr>
</thead>
<tbody>
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<td>27311</td>
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<td>19949</td>
<td>37.97</td>
<td>4188</td>
<td>7.97</td>
</tr>
<tr>
<td>3</td>
<td>15538</td>
<td>14229</td>
<td>91.58</td>
<td>903</td>
<td>5.81</td>
<td>104</td>
<td>0.67</td>
</tr>
<tr>
<td>5</td>
<td>52901</td>
<td>39382</td>
<td>74.44</td>
<td>10274</td>
<td>19.42</td>
<td>2929</td>
<td>5.54</td>
</tr>
<tr>
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<td>4146</td>
<td>32.54</td>
<td>4774</td>
<td>37.47</td>
<td>2419</td>
<td>18.98</td>
</tr>
<tr>
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<td>n.s.</td>
<td>n.s.</td>
<td>n.s.</td>
<td>n.s.</td>
<td>n.s.</td>
<td>n.s.</td>
</tr>
<tr>
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<td>107234</td>
<td>97.08</td>
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<td>306</td>
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<td>154</td>
<td>0.44</td>
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<td>11008</td>
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<td>409</td>
<td>2.45</td>
</tr>
<tr>
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<td>5601</td>
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</tr>
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<td>47976</td>
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<td>17.26</td>
<td>4372</td>
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</tr>
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<td>61.13</td>
<td>7539</td>
<td>30.57</td>
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</tr>
<tr>
<td>Total</td>
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<td>86766</td>
<td>13.83</td>
<td>22081</td>
<td>3.52</td>
</tr>
</tbody>
</table>

In part, the number of probes is lower than the number of primary screening of previous tables (marked with "a").
Figure 2: Age at blood collection 2005 to 2012

Figure 3: Period between sampling and laboratory receipt 2005 to 2012

Figure 4: Period from laboratory receipt to report 2005 to 2012
5 Time of screening in the confirmed cases

5.1 Primary screening

Crucial for a successful screening is the reliability of results and the promptness of further diagnostic evaluation and therapy in suspect cases. The optimal sampling time is the 48th to the 72nd hour of life. The probe should not be sampled before the 36th and not after the 72nd hour of life. Any delay means a potential risk for affected children.

The time of primary screening is shown for the targeted disease in Table 5.1. For clarity reasons the description >72 hours of age is reported in days. About 6.9% of diseased children were at the time of sampling older than 72 hours.

Table 5.1 Time of primary screening in confirmed cases

<table>
<thead>
<tr>
<th>Disease</th>
<th>36-72h</th>
<th>4-7d</th>
<th>&gt;7d</th>
<th>&lt;36h</th>
<th>&lt;32WOG</th>
<th>≥36h, Time not specified*</th>
<th>Not specified**</th>
<th>Total</th>
</tr>
</thead>
<tbody>
<tr>
<td>Hypothyroidism</td>
<td>171</td>
<td>13</td>
<td>5</td>
<td>16</td>
<td></td>
<td></td>
<td></td>
<td>205</td>
</tr>
<tr>
<td>CAH</td>
<td>37</td>
<td>2</td>
<td>7</td>
<td>1</td>
<td>1</td>
<td></td>
<td></td>
<td>48</td>
</tr>
<tr>
<td>Biotinidase</td>
<td>36</td>
<td>1</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>37</td>
</tr>
<tr>
<td>Classic Galactosaemia</td>
<td>9</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>9</td>
</tr>
<tr>
<td>PKU/HPA</td>
<td>109</td>
<td>11</td>
<td>1</td>
<td>4</td>
<td>1</td>
<td></td>
<td></td>
<td>126</td>
</tr>
<tr>
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<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>5</td>
</tr>
<tr>
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<td>3</td>
<td>1</td>
<td></td>
<td></td>
<td>62</td>
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<td></td>
<td>2</td>
<td></td>
<td></td>
<td></td>
<td>7</td>
</tr>
<tr>
<td>VLCAD</td>
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<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>7</td>
</tr>
<tr>
<td>GA I</td>
<td>5</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>5</td>
</tr>
<tr>
<td>IVA</td>
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<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>5</td>
</tr>
<tr>
<td><strong>Total</strong></td>
<td>438</td>
<td>34</td>
<td>2</td>
<td>19</td>
<td>20</td>
<td>2</td>
<td></td>
<td>516</td>
</tr>
</tbody>
</table>

* ≥36h Not specified does not include repeat testing with early sampling or preterm birth, exact age of sampling time not stated.
** No information, neither WoG nor age at sampling.
5.2 Result of the primary testing and time of sampling in the confirmed cases

We stratified in the confirmed cases the results of the primary test card according to sampling time and result. For premature infants for whom the test was performed before the 32nd week of gestation we itemised the sufficiency of the control algorithm. In two cases of hypothyroidism the control beyond 32nd week of gestation showed a normal result. These children were diagnosed later. In 3 children the compulsory screening control was not undertaken timely. It is not known if and how many premature infants below the 32nd week of gestation with congenital hypothyroidism have been undiagnosed due to unremarkable secondary or missed screening.

Table 5.2 : Confirmed cases: Result of the primary testing and time of sampling

<table>
<thead>
<tr>
<th>Disease</th>
<th>Full term, sampling ≥36h</th>
<th>Sampling &lt;36h</th>
<th>First sampling &lt;32 WOG</th>
<th>Not specified</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Prim. screen suspicious</td>
<td>Prim. screen suspicious</td>
<td>Prim. screen suspicious, screening at 32 WOG suspicious</td>
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</tr>
<tr>
<td></td>
<td>Prim. screen not suspicious</td>
<td>Prim. screen not suspicious</td>
<td>Prim. screen not suspicious, missed screening at 32 WOG</td>
<td>Prim. screen not suspicious, screening at 32 WOG</td>
</tr>
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<td>Hypothyroidism</td>
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<td>7</td>
<td>7 3 2</td>
<td>1</td>
</tr>
<tr>
<td>CAH</td>
<td>38 1 a 7 1 1</td>
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<td></td>
<td></td>
</tr>
<tr>
<td>Biotinidase</td>
<td>37</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Classic Galactosaemia</td>
<td>9</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>PKU/HPA</td>
<td>121 4</td>
<td></td>
<td></td>
<td>1</td>
</tr>
<tr>
<td>MSUD</td>
<td>5</td>
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<td></td>
</tr>
<tr>
<td>MCAD</td>
<td>58 3 1</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>LCHAD</td>
<td>5 2</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>VLCAD</td>
<td>7</td>
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<td></td>
<td></td>
</tr>
<tr>
<td>GA I</td>
<td>5</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
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<td></td>
<td></td>
</tr>
<tr>
<td>Total</td>
<td>474 1 19 8 7 3 2 2</td>
<td></td>
<td></td>
<td></td>
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</tbody>
</table>

a Mutation: I172N

6 Confirmation of pathological results

The following chapter outlines the diagnostic measures for confirmation of the diagnosis, as known to the laboratories. This information is used for quality control by the individual laboratories but does not always get reported by the physicians taking care of the patient. For the year 2012, 24 out of 516 confirmed cases had no detailed information about the confirmation diagnostics available, the available data though allows a plausible analysis. In a further 22 cases only limited information is given that confirmation can not be accepted and we therefore do not list it in our analysis.
### 6.1 Hypothyroidism

<table>
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<tr>
<th>Lab</th>
<th>Confirmed cases*</th>
<th>TSH (Serum)</th>
<th>T3</th>
<th>fT3</th>
<th>T4</th>
<th>fT4</th>
<th>Ultrasound</th>
<th>Thyroid antibodies</th>
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<td>18</td>
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<td>16</td>
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<td>7</td>
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<td>3</td>
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<td>7</td>
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<tr>
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<td>14</td>
</tr>
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<tr>
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<tr>
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</tr>
<tr>
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* incl n=4 cases without detailed information of confirmation diagnostics

### 6.2 Congenital adrenal hyperplasia (CAH)

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<th>Urinary steroids</th>
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* incl n=2 cases without detailed information of confirmation diagnostics
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* incl n=1 cases without detailed information of confirmation diagnostics

### 6.4 Classic Galactosaemia

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* incl n=1 cases without detailed information of confirmation diagnostics

### 6.5 PKU / HPA

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* incl n=3 cases without detailed information of confirmation diagnostics
### 6.6 MSUD

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### 6.7 MCAD-Deficiency

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* incl n=13 cases without detailed information of confirmation diagnostics

### 6.8 LCHAD-Deficiency

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### 6.10 No confirmed cases of CPT I-Deficiency, CPT II-Deficiency and CACT-Deficiency

### 6.11 Glutaric aciduria Type I

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<th>Molecular genetic testing</th>
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### 6.12 Isovaleric acidaemia

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7 Methods and cut offs in screening

7.1 Filter paper for sampling

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7.2 Hypothyroidism

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<td>3</td>
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* Full term and premature babies – at elevated Delfia 17OHP TMS Steroid profile with 17OHP, 21-Desoxycortisol, 11-Desoxycortisol, Cortisol und Androstendion.

### 7.4 Biotinidase deficiency

<table>
<thead>
<tr>
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<th>Parameter</th>
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<th>Method</th>
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<tbody>
<tr>
<td>1</td>
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</tr>
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<td>8</td>
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<td>30% daily mean</td>
<td>Colorimetrie quantitative</td>
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<tr>
<td>9</td>
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<td>Colorimetrie qualitative</td>
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<td>Colorimetrie qualitative</td>
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<td>Biotinidase</td>
<td>&lt; 30%</td>
<td>Fluometrie quantitative</td>
</tr>
<tr>
<td>13</td>
<td>Biotinidase</td>
<td>&lt; 30%</td>
<td>Fluometrie quantitative</td>
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<tr>
<td>14</td>
<td>Biotinidase</td>
<td>&lt; 30%</td>
<td>Colorimetrie quantitative</td>
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<tr>
<td>15</td>
<td>Biotinidase</td>
<td>&lt; 30%</td>
<td>Colorimetrie quantitative</td>
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### 7.5 Galactosaemia

<table>
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<td>GALT</td>
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<td>BIORAD Quantase</td>
</tr>
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<td>15 mg/dl</td>
<td></td>
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<tr>
<td>6</td>
<td>GALT</td>
<td>3,5 U/gHb</td>
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<tr>
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<td>Galactose</td>
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<td>20 mg/dl</td>
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<tr>
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<td>BIORAD Quantase</td>
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<tr>
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<td>GALT</td>
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<td>BIORAD Quantase</td>
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<tr>
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<td>Galactose</td>
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<td>BIORAD Quantase</td>
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* Galactose as a second-tier method

### 7.6 MS/MS

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Literature
