The results of karyotyping for Down’s syndrome in neonates were surveyed. From local data 36%, and from a national questionnaire, 32% of such samples were negative for Down’s syndrome. To examine this, a subset of notes was reviewed for documentation of clinical signs of Down’s syndrome. Some characteristics were often recorded, but other common discriminatory characteristics were noted less often or not at all.

The clinical diagnosis of Down’s syndrome in the neonatal period has been described as “seldom a problem to the neonatologist”, but the variable nature of the presenting features is such that the diagnosis can be uncertain. When the diagnosis is suspected, it is good practice to inform the parents and await confirmation of the karyotype, which can take two or three days. This wait for confirmation of a life changing diagnosis is one of great anxiety for parents. Maximising clinical diagnostic accuracy is therefore important, and several studies documenting the most characteristic physical features including diagnostic indices have been published, but only two of these have looked specifically at the diagnosis of Down’s syndrome in the newborn.

The purpose of this study was to survey the results of karyotyping for Down’s syndrome in neonates locally and nationally, and to investigate which criteria are being used to reach a diagnostic suspicion of Down’s syndrome.

METHOD
Two cytogenetic laboratories in Greater Manchester (Royal Manchester Children’s Hospital and St Mary’s Hospital) performed database searches to identify all samples of babies aged 0–4 weeks analysed for a diagnostic query of Down’s syndrome for the period 1 January 1999 to 30 December 2000. These laboratories process samples from several paediatric departments in Greater Manchester and North West England. These data were then used to determine those infants with karyotypes not compatible with Down’s syndrome (Down’s syndrome negative, DSN) and those who had a karyotype consistent with Down’s syndrome (Down’s syndrome positive, DSP).

A questionnaire was then sent to 25 other cytogenetic laboratories in the United Kingdom asking for information about the number of requests processed in the last two years for a diagnostic index being used.

RESULTS
The two Manchester centres processed 174 cases that met the above criteria. These requests were received from 27 hospitals. Of these, 109 were DSP and 63 were DSN. Thus 36% of the cases referred for cytogenic testing were not Down’s syndrome.

Of the 29 factors included on the proforma, five are routinely available from a standard newborn examination (maternal age, birth weight, presence or absence of jaundice, admission to special care, feeding difficulties), and were documented for all 72 babies. The head circumference was noted in 89% of the 72 babies, a wide gap between 1st and 2nd toes in 80%, and a flat nasal bridge in 57%. Reference was made to determine how often, if ever, this situation arises. When the diagnosis is considered possible, the threshold for performing karyotype testing and informing the parents is reached. Our results suggest that, where this threshold is reached, there is about a two thirds chance of the baby having Down’s syndrome.

DISCUSSION
Midwifery staff usually express the initial concern that a baby has Down’s syndrome. Clinical examination can often confirm or refute this with confidence. If there are no clinical grounds for making the diagnosis, the parents can reasonably be kept unaware of the initial suspicion; this study has not been able to determine how often, if ever, this situation arises. When the diagnosis is considered possible, the threshold for performing karyotype testing and informing the parents is reached. Our results suggest that, where this threshold is reached, there is about a two thirds chance of the baby having Down’s syndrome.

Abbreviations: DSN, Down’s syndrome negative; DSP, Down’s syndrome positive
syndrome. Part of the reason for this high DSN rate may be that some samples are sent after delivery to allay parental anxiety following antenatal serum screening results in the absence of significant dysmorphism (10/41 DSN cases from one centre, although this reason was not apparent in the 72 sets of notes we reviewed locally).

We found no recent studies detailing the clinical diagnostic accuracy of Down’s syndrome in the newborn. Fried, using a diagnostic index, was able to accurately identify the diagnosis on clinical grounds for 22 of 30 (73%) newborn babies suspected of having Down’s syndrome. This value is by no means directly comparable with the results of our study because the Fried study used a prescribed set of physical characteristics in babies examined prospectively by a single interested clinician. Our study reflects a current working view of the issue, with data coming from a broad cohort of paediatricians.

The retrospective study of notes has limitations because what is recorded may not accurately reflect how decisions were reached. Nevertheless, the review indicates that diagnostic indices do not appear to have been adopted. Also it appears that many important traits are not often recorded as contributing to a diagnostic decision, whereas well known but relatively poorly discriminating characteristics commonly figure. Of the three physical features most often recorded in the notes review—that is, hypotonia, slanting palpebral fissures, and Simian crease—the latter two are relatively non-specific. Excess neck fat pad, an easily noted and more specific feature, was recorded much less often, as were other important signs (table 1). Other less well known but useful diagnostic features such as dermatoglyphics, ear length, the pursed eyelids, and vertical wrinkling sign, and the Smithells hypotonia test were not recorded.

There have been, to our knowledge, no studies looking at the effects of a false positive diagnosis of Down’s syndrome on parents, but common sense suggests that such a suspected diagnosis should be discussed on as informed a basis as possible. (One of the authors has direct experience of such a scenario where the parents still felt resentful one year later about the false diagnosis and did feel that it had marred their early enjoyment of their newborn daughter.) A targeted and systematic examination of the baby using simply observed external physical characteristics (table 2) may allow a more informed discussion to take place with parents, including a more accurate weighting of the likelihood or otherwise of a positive diagnosis.

<table>
<thead>
<tr>
<th>Characteristic</th>
<th>Percentage recorded in current study (n=72)</th>
<th>Most discriminatory characteristic 1–8 (Fried3)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Excess neck skin</td>
<td>47</td>
<td>1</td>
</tr>
<tr>
<td>Mouth corners turned down</td>
<td>0</td>
<td>2</td>
</tr>
<tr>
<td>Hypotonia</td>
<td>100</td>
<td>3</td>
</tr>
<tr>
<td>Flat face</td>
<td>57</td>
<td>4</td>
</tr>
<tr>
<td>Dysplastic ear</td>
<td>89</td>
<td>5</td>
</tr>
<tr>
<td>Epicanthic fold</td>
<td>Data not collected</td>
<td>6</td>
</tr>
<tr>
<td>Gap 1st/2nd toes</td>
<td>80</td>
<td>7</td>
</tr>
<tr>
<td>Protruding tongue</td>
<td>24</td>
<td>8</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Characteristics</th>
<th>Number of characteristics</th>
<th>Conclusion</th>
<th>DSN</th>
<th>Unclear</th>
<th>DSP</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>0–2</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>3–5</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>6–8</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Error</td>
<td>No false +ves</td>
<td>No false +ves (&lt;1/100 DSP babies)</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>No false –ves</td>
<td>No false +ves (&lt;1/100000 DSN babies)</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

DSN, Down’s syndrome negative; DSP, Down’s syndrome positive.

References

Diagnosis of Down's syndrome in neonates

D Hindley and S Medakkar

Arch Dis Child Fetal Neonatal Ed 2002 87: F220-F221
doi: 10.1136/fn.87.3.F220

Updated information and services can be found at:
http://fn.bmj.com/content/87/3/F220

These include:

References
This article cites 3 articles, 1 of which you can access for free at:
http://fn.bmj.com/content/87/3/F220#BIBL

Email alerting service
Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

Notes

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/