EARLY DIAGNOSIS AND EARLY THERAPY IN
CONGENITAL CRETINISM

BY

Y. ÅKERRÉN

From the Medical Department, Children's Hospital, Gothenburg, Sweden

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How soon can the diagnosis of congenital cretinism be made? Isolated cases have been diagnosed at birth (Abels, 1911). There is a photograph in Potter's (1952) monograph showing an infant at the age of 1 month with the typical facies; the case had been diagnosed and, judging by a later view, successfully treated by McIntosh (1952). Wieland (1940), who has great experience, claims that he observed six or eight cases aged between 3 and 5 months, where the diagnosis was obvious at first sight. All these patients had previously been under the observation of other physicians for symptoms which definitely or very probably were connected with congenital cretinism. Wieland's most successful case, from the therapeutic point of view, was diagnosed and treated from the age of 5 months.

According to Higgins and Ingalls (1948) 'the first signs of cretinism will appear at the age of about 2 months, although the condition is seldom diagnosed before the child is 5 months old'. Many larger series, especially of an early date, contain only few with an early diagnosis and therapy, e.g. Marvel's (1939) from Oslo. A fairly large series from Sweden (d'Avignon and Melin, 1949) with 22 cases includes 10 with a diagnosis made before the age of 5 months.

Why is the diagnosis so often made so late? Frequently the parents do not consult the physician early enough. Another and usual reason is that the physician does not recognize the picture of the disease at its early stage of development. This certainly also applies to paediatricians with great experience. Cases of congenital cretinism are so rare that only one or two per annum are diagnosed at the larger children's hospitals in Scandinavia. Finally, there are cases of mild congenital cretinism where the typical symptoms do not develop until later on, at the end of the first year of life or later.

In a previous paper (Åkerrén, 1954) I have been able to point out the frequent coincidence between congenital cretinism and icterus neonatorum prolonged with a remarkable duration. In this investigation I combined 10 cases of congenital cretinism which also had shown signs of icterus with a duration of at least six weeks, but otherwise, as far as could be judged, of a physiological type. At an investigation carried out at the Children's Hospital of Gothenburg, including 946 newborn infants who were closely studied for the occurrence and duration of physiological icterus, Beskow, among others, found that the mean duration of icterus which lasted at least 48 hours was 11.37 ± 6.20. A duration of physiological icterus exceeding about 30 days must thus be very rare.

As shown in Table 1, early diagnosed cases of congenital cretinism are unusual. In the table all the cases are given which were admitted to the Children's Hospital of Gothenburg before the age of 1 year, from 1922 up to and inclusive of the spring of 1954. They are in all 18. Of the nine babies admitted before they had reached the age of 3 months, no fewer than six had shown icterus with a duration of at least six weeks. Of the five babies

<p>| TABLE 1 |</p>
<table>
<thead>
<tr>
<th>CASES WITH CONGENITAL MYXOEDEMA ADMITTED TO THE CHILDREN'S HOSPITAL, GOTHENBURG, BEFORE THE AGE OF 1 YEAR DURING 1922-34</th>
</tr>
</thead>
<tbody>
<tr>
<td>Group 1</td>
</tr>
<tr>
<td>Cases Admitted</td>
</tr>
<tr>
<td>Cases with prolonged icterus</td>
</tr>
<tr>
<td>Cases without any statement of icterus</td>
</tr>
<tr>
<td>Total</td>
</tr>
</tbody>
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* Had icterus to the age of 2½ months.
admitted between the ages of 3 and 5 months, one
had suffered from icterus up to the age of 2½ months.

The coincidence of comparatively unusual con-
ditions, such as early diagnosed congenital cretinism
and icterus of a remarkably long duration is not
likely to be due to chance.

This coincidence has been mentioned in a few
case reports but otherwise not observed or com-
cmented upon. The reason seems to be that even
experienced paediatricians only rarely get an
opportunity of seeing and diagnosing early cases of
congenital cretinism. An infant who during the
first months of life shows icterus neonatorum of
remarkable duration should thus be carefully
observed and examined until the diagnosis can be
made or excluded with certainty.

Practically all textbooks and handbooks of paediat-
rics unanimously point out that an early diagnosis
and adequate treatment are of greatest importance in
the prognosis of mental development in congenital
cretinism. The prognosis for the somatic symptoms
is, generally, favourable even if the diagnosis
was delayed. These results have been indicated
in Topper's series, which the final results
regarding the mental development was 'defective',
there being a psychosis present, but where the I.Q.
was high.

The differences between the frequency of an I.Q.
of less than 80 in the various groups has been
statistically analysed by means of the $\chi^2$ method.
Between groups 1 and 2 there is a difference
significant at the 5% level. Between groups 1 and 3
and 2 and 3 there is no statistically significant
difference.

Judging by the statistical study of the series
combined here, it seems justifiable to conclude that
treatment started before the age of 5 months gives
more hope of a favourable result than treatment
begun later on. It may seem remarkable that such a
comparatively large number of the results obtained
in group 3 are favourable, although these results
do not differ in a statistically significant way from
those obtained in group 2. This is probably due to
the fact that this group is likely to be less homo-
genous than the other two. Thus it includes some
severe cases where the therapy was begun late with
a bad result as a sequel, but it probably also includes
some slight cases in which it was impossible to make
the diagnosis earlier. In these cases the insufficiency
of thyroid has not been severe. The central nervous
system and especially the brain have not been
subjected to such early damage from or early after birth as when there is an already severely deficient thyroid function. The results have, therefore, to rather a large extent been successful.

Table 3 illustrates, although it is not sufficiently extensive for statistical studies, the differences in results between the three groups. The table shows that the most severe disturbances with I.Q. below 30 only occur in group 2 and that in no case where the treatment was begun before the age of 5 months was an I.Q. below 51 found.

### Summary

There is an obvious coincidence between congenital cretinism and icterus neonatorum of excessive duration. In babies with congenital cretinism who come under observation before the age of 3 months, icterus seems to be very frequent.

The knowledge of this syndrome must be assumed to be of practical importance for the early diagnosis of congenital cretinism. In cases of icterus neonatorum of excessive duration careful observation and examination must be performed until the diagnosis can either be made or excluded.

In cases of severe congenital cretinism early treatment, i.e., beginning before the approximate age of 5 months, gives a much greater chance of obtaining a favourable mental prognosis than if the treatment is started later.

As an early diagnosis of congenital cretinism is a reason for early adequate therapy, the knowledge of the congenital cretinism-icterus neonatorum syndrome is of value from the therapeutic and prognostic points of view.

### References


McIntosh, R. (1952). Quoted from Potter, p. 278.


