Rubella Retinopathy

An account of six cases

GORDON STARK

From the Royal Hospital for Sick Children, Sciennes Road, Edinburgh

The pigmentary retinopathy which may follow prenatal rubella is not rare and has been well documented in the ophthalmological literature. Gregg attributed the first observation of it to Aileen Mitchell who, in 1939, reported a case of left monocular cataract and stated 'the fundus of the right eye appeared pale and some scattered irregular spots of pigment were observed' (Gregg, 1941; 1946). The early Australian reports were followed by others from the U.S.A., South America, and Europe (Reese, 1944; Ramon-Guerra, 1955; Franceschetti and Bourquin, 1946). Thus, within five years of Gregg's original description of the rubella syndrome, the retinal lesion was well known to ophthalmologists. It is, however, still not widely known among paediatricians and was not mentioned among a variety of minor abnormalities in the Ministry of Health's large prospective study of children born following pregnancy complicated by rubella in the first 16 weeks (Sheridan, 1964). Nevertheless, without a special search, six cases have been encountered in less than a year in the Neurological Unit of the Royal Hospital for Sick Children, Edinburgh. Since recognition of rubella retinopathy may be of diagnostic and prognostic value, it seemed worth while drawing attention to it, and to this end these cases will be outlined and the condition discussed.

Clinical Findings

From Table I which summarizes the case histories of patients it can be seen that in Cases 1, 2, 3, and 4, the maternal illness was diagnosed at the time as typical rubella. In Case 5, the occurrence of cataract and in Case 6, deafness, suggested that in these, too, the mother had suffered from rubella rather than measles.

The typical appearance which we have seen and which is recorded in the literature is illustrated in Fig. 1. There is abnormal pigmentation concentrated at the posterior pole of the eye and consisting of sprinkled dust-like black particles interspersed with larger dots and

Received November 22, 1965.

<table>
<thead>
<tr>
<th>Case No.</th>
<th>Sex and Age (yr.)</th>
<th>Family History of Congenital Malformations</th>
<th>Pregnancy</th>
<th>Labour and Delivery</th>
<th>Birth Wt (kg.)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>F 4½</td>
<td>None</td>
<td>Rubella 9th week, despite y-globulin 5 days</td>
<td>Normal</td>
<td>2.4</td>
</tr>
<tr>
<td>2</td>
<td>F 2½</td>
<td>Mother had previous miscarriage, stillborn</td>
<td>Rubella at 9 weeks</td>
<td>Normal</td>
<td>2.7</td>
</tr>
<tr>
<td>3</td>
<td>M 2½</td>
<td>None</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>4</td>
<td>M 7</td>
<td>Mongol sib</td>
<td>Rubella 4th month</td>
<td>Normal</td>
<td>3.0</td>
</tr>
<tr>
<td>5</td>
<td>M 1½</td>
<td>None</td>
<td>Rubella or measles at end of 1st month; influenza 2 weeks later</td>
<td>Normal</td>
<td>3.3</td>
</tr>
<tr>
<td>6</td>
<td>M 7½</td>
<td>None</td>
<td>Measles or rubella 3rd month</td>
<td>Normal</td>
<td>3.6</td>
</tr>
</tbody>
</table>

TAB2.

Summrv
Rubella Retinopathy

The finding of this pigmentary retinopathy which is a common component of the rubella syndrome may shed light on the aetiopathology of associated congenital malformations if there is no history of maternal rubella, or if, as is not infrequent, the history is of a nondescript illness or skin rash in early pregnancy. The appearance and central distribution of the lesion are sufficiently characteristic to distinguish it from retinitis pigmentosa which may also be associated with deafness and neurological lesions and in which the prognosis for vision is poor. The presence of atrophy of the iris stroma, though not seen in our cases, may confirm the diagnosis of rubella retinopathy (Emerson, 1958).

The high incidence of neurological disorders in the patients described here is probably the result of selection and has not been described in other reports of the rubella syndrome with or without retinopathy. By far the most commonly associated lesion is nerve deafness which was present in all but one of our cases, and is outstanding in those reported in the literature (Table II). The pre-eminence of deafness as an associated lesion is arresting, and the fact that it tends to follow infection relatively late in the first trimester (the cochlea first appears in the 7th week) suggests that retinopathy may also result from

Cases

<table>
<thead>
<tr>
<th>Condition at Birth</th>
<th>Fundi</th>
<th>Lesions Associated With Retinopathy</th>
<th>IQ</th>
<th>EEG</th>
</tr>
</thead>
<tbody>
<tr>
<td>Good</td>
<td>Pigmentation at posterior poles and midzone</td>
<td>Deafness; ataxia</td>
<td>85</td>
<td>Normal</td>
</tr>
<tr>
<td>Good</td>
<td>Pigmentation at posterior poles</td>
<td>Deafness; ataxia</td>
<td>Low 70's</td>
<td>Normal</td>
</tr>
<tr>
<td>Good</td>
<td>Pigmentation at posterior poles left &gt; right</td>
<td>Deafness</td>
<td>Above average</td>
<td>Normal</td>
</tr>
<tr>
<td>Good</td>
<td>Pigmentation at posterior poles and midzone</td>
<td>Deafness; mild diplegia</td>
<td>90-95</td>
<td>Normal</td>
</tr>
<tr>
<td>Good</td>
<td>Pigmentation at posterior poles</td>
<td>Bilateral cataracts</td>
<td>Mild developmental retardation at 15 months &lt; 50</td>
<td>Normal (EEG, audiometry also normal)</td>
</tr>
<tr>
<td>Apnoeic, resuscitated with difficulty</td>
<td>Pigmentation at posterior poles</td>
<td>Deafness; diplegia; mental defect</td>
<td></td>
<td>Normal</td>
</tr>
</tbody>
</table>

Fig. 1.—Typical rubella retinopathy. (Reproduced by kind permission of the publishers of Ophthalmologica.) (Franceschetti, Dieterle, and Schwarz, 1958).
TABLE II
Lesions Reported in Association with Retinopathy

<table>
<thead>
<tr>
<th>Lesions</th>
<th>No.</th>
</tr>
</thead>
<tbody>
<tr>
<td>(A) Cataract</td>
<td>11</td>
</tr>
<tr>
<td>(B) Cataract + congenital heart disease</td>
<td>5</td>
</tr>
<tr>
<td>(C) Cataract + congenital heart + deafness</td>
<td>1</td>
</tr>
<tr>
<td>(D) Congenital heart disease</td>
<td>1</td>
</tr>
<tr>
<td>(E) Congenital heart disease + deafness</td>
<td>75</td>
</tr>
</tbody>
</table>

94

comparatively late infection. The majority of reported cases of retinopathy with deafness, however, come from three large ophthalmological surveys of rubella-deafened children in which unfortunately the timing of maternal infection is not stated (Marks, 1946; Hamilton, Phillips, Palfreyman, and Waterworth, 1948; Morlet, 1949). An attempt is made in Fig. 2 to summarize reported cases (including the 6 described) which are adequately documented as regards both the associated lesions and the timing of maternal infection. In the first month, cataract alone, or accompanied by congenital heart disease, is the main associated lesion, while in the second and third months deafness takes over. It is evident also that these cases of retinopathy are evenly spread over the first three months with respectively 9, 9, and 10 cases in each. The evenness of this distribution over the first three months, which contrasts strikingly with the marked tendency for the incidence of the other lesions to fall off, as shown, for example, by Dekaban, O'Rourke, and Cornman (1958), has embryological implications which call for brief consideration.

Pigment begins to appear in the outer layer of the optic cup at the 7 mm. stage, and has increased considerably by the 10 mm. stage, placing the important period of pigment deposition in the 5th week of embryonic life. This is earlier than the development of even the primary lens fibres, which begin in the 6th week (Mann, 1964). It is, therefore, somewhat surprising that though cataract has only rarely been reported with deafness, the combination of retinopathy and deafness, a relatively late lesion, is common. The possibility arises, therefore, that the rubella virus may act not only by interfering

Fig. 2.—Associated lesions and timing of maternal infection in 28 cases of rubella retinopathy. This figure is based on present cases and the following (Babel and Dieterle, 1980; Beswick, Warner, and Warkany, 1949; Blankstein and Feiman, 1952; Cordes, 1945; Dekaban et al., 1958; Emerson, 1958; Franceschetti and Bourguin, 1946; Francois and Verriest, 1956; Gregersen, 1958; Hopkins, 1949; Ellett, 1945; Long and Danielson, 1945; Swan, Tostevin, Moore, Mayo, and Black, 1943; Swan, Tostevin, and Black, 1946).
Rubella Retinopathy

with developing pigment epithelium, but at least in some of the later cases by damaging that already formed. In this context, a case described by Lundström and Boström (1958) of irregular fundal pigmentation in a child whose mother had rubella in the 8th month of pregnancy is interesting. Moreover, the retinopathy may occasionally be simulated by retinal changes resulting from postnatal viral infections, including measles (Walsh, 1947) and mumps (Bischler, 1945).

While it is generally true that the lesions found in the rubella syndrome depend on the stage of prenatal development at the time of infection, the resultant malformations are less closely related to the timetable of embryonic development than those following, for example, thalidomide administration. In addition to the possibility suggested above that later infection may damage tissues that have developed earlier, it may be, conversely, that organs that have not even begun to develop at the time of maternal infection are malformed as a result of persistence of the virus in the embryo; this has recently been demonstrated (Kay, Peppercorn, Porterfield, McCarthy, and Taylor-Robinson, 1964). This would explain, for instance, the not unusual cases of deafness following maternal rubella in the first month and cases such as the child described by Dekaban et al. (1958), whose mother had rubella in the fourth week of pregnancy and who showed no fewer than 11 congenital abnormalities, including deafness, cataract, heart disease, and retinopathy.

Summary and Conclusions

Six cases of rubella retinopathy seen within a year in a children’s hospital are described and the condition is discussed.

Unlike retinitis pigmentosa, rubella retinopathy is not progressive and has little, if any, effect on vision. Its main significance is its ability to ‘label’ other congenital malformations such as cataract, heart disease, and deafness in the absence of a definite history of maternal rubella. This, and the differential diagnosis from retinitis pigmentosa, make its recognition of importance to paediatricians.

I am grateful to Dr. T. T. S. Ingram for permission to publish these cases, and for his encouragement and helpful criticism during the preparation of this report. I am indebted to Messrs. S. Karger, Basel/New York, for permission to reproduce Fig. 1.

References


Cordes, F. C. (1945). In Discussion to paper by Goar and Potts (see above), p. 142.
Elliott, E. C. (1945). In Discussion to paper by Goar and Potts (see above), p. 145.
——— (1946). In Discussion to paper by Marks, p. 124.
Morlet, C. (1949). Rubella retinitis in Western Australia. ibid., 9, 212.
Rubella Retinopathy: An Account of Six Cases

Gordon Stark

Arch Dis Child 1966 41: 420-423
doi: 10.1136/adc.41.218.420

Updated information and services can be found at:
http://adc.bmj.com/content/41/218/420.citation

Email alerting service

These include:
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/