CASE REPORT

ANIRIDIA: A RARE MANIFESTATION OF CONGENITAL RUBELLA SYNDROME

Ali Shabbir Hussain, Syed Rehan Ali, Nadia Mohammad, Nabiha Ali, Shakeel Ahmed, Tauseef Ahmad*

Department of Pediatrics and Child Health, Aga Khan University, Stadium Road, Karachi, *Ayub Medical College Abbottabad-Pakistan

A foetus affected by a congenital rubella infection can develop congenital rubella syndrome (CRS). Aniridia is the absence of iris, rarely been described in literature in association with CRS, can easily be overlooked, leading to complications e.g. glaucoma and blindness later in life. We report a case of a neonate with CRS and aniridia presenting at a tertiary care hospital.

Keywords: Aniridia; Congenital; Rubella; Syndrome; Iris; Hypoplasia

Citation: Ali R, Hussain AS, Mohammad N, Ali N, Ahmed S, Ahmad T *et al*. Aniridia: A rare manifestation of Congenital Rubella Syndrome. J Ayub Med Coll Abbottabad 2019;31(1):131–3.

INTRODUCTION

The foetus acquires rubella through the course of pregnancy. The clinical significance depends on the time of the mother's acquisition of the virus. If maternal infection happens to be in the first trimester, the risk of rubella-associated defects is greatly increased. According to Pumper and Yamashiroya, between 50% and 80% of foetuses exposed to maternal rubella virus become infected prior to the 8th weeks of gestation. Infection of the foetus is fairly uncommon, during the 3rd trimester which is 6–10%. The eyes, the ears, the heart, the central nervous system and the brain appear to be especially susceptible to rubella-associated damage.[1] Aniridia is the absence of iris, has rarely been described in literature in association with CRS. It is a rare association which can easily be overlooked, and can lead to complications e.g. glaucoma and blindness later in life. We report the case of a neonate with CRS and aniridia presenting at a tertiary care hospital – the first of its kind reported in literature.

CASE PRESENTATION

A female, full term neonate, weighing 2.4 kg, with symmetrical Intra uterine growth retardation (IUGR) was born to a young primigravida mother at a primary care hospital. The baby was referred to our hospital on the second day of life for management of cardiac failure secondary to Patent Ductus Arteriosus (PDA). Her antenatal course had been uneventful. The third trimester foetal ultrasound depicted IUGR. The mother had not been previously vaccinated against rubella, nor had she been investigated for antibodies against rubella during the pregnancy. She had not noticed any clinical manifestations of rubella during the pregnancy.

On clinical examination, all anthropometric measurements were below the tenth percentile. The baby was in obvious respiratory distress and required oxygen supplementation at a rate of 2 litters/minute. Her heart rate was 180 beats/minute and her blood pressure was 60/40 mm Hg. A harsh, grade 4/6 systolic murmur was heard at the left 1st intercostal space. Eye examination showed bilateral aniridia with clear media. No corneal oedema was noted. Posterior segment examination showed a normal looking optic disc and retina. Intra-ocular pressure was 18 mm Hg. Anterior segment photographs were taken (Figure-1, 2). No red reflex was detected on indirect ophthalmoscopy.

The baby's Complete Blood Count (CBC) was within normal limits. Patient's cytogenetic work-up was negative for trisomy 13, 18 and 21; however, we did not have Fluorescent in Situ Hybridization (FISH) available to check for specific micro deletions. Echocardiography showed PDA. A probable diagnosis of congenital rubella syndrome was made and serum samples of the baby and her mother were sent to check for rubella antibody titres. Her serum IgM for Rubella virus was positive; hence the diagnosis of CRS was confirmed. Appropriate treatment was initiated and the child is being followed up. Her next visit will be at the age of 6 months, when her condition will be reviewed. She will also undergo auditory examination at that time.

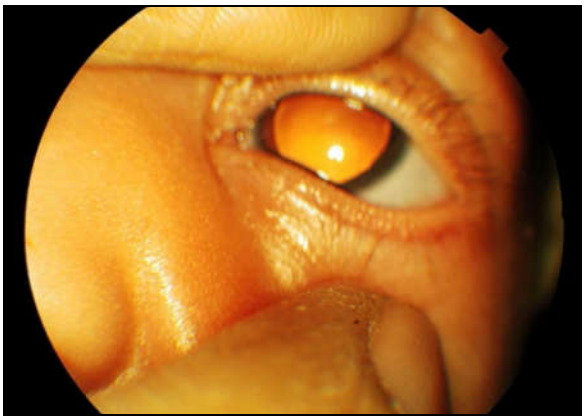

Figure-1: Photograph of the left eye of a neonate presenting with bilateral aniridia as a manifestation of congenital rubella syndrome.

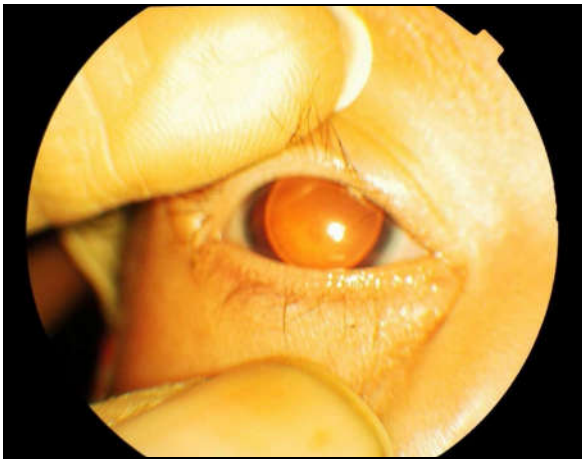

Figure-2: Photograph of the right eye of a neonate presenting with bilateral aniridia as a manifestation of congenital rubella syndrome.

DISCUSSION

Congenital rubella syndrome (CRS) was first described by an ophthalmologist in 1941, who found an association of cataract in a new-born whose mother had German measles during pregnancy.[2] He also described the atrophic appearance of the iris in a few cases.[2] Congenital rubella syndrome presents in new-borns as an amalgam of blindness, deafness, microcephaly, mental retardation and cardiac defects. It can easily be prevented by timely immunization to all reproductive age girls. According to world health organization (W.H.O) there are almost 100,000 neonates affected worldwide annually due to CRS.[3]

Congenital rubella syndrome affects almost all ocular structures, either in isolation or in combination. Ocular findings occur in 43% of children, reported in different studies.[3,4] Rubella cataract is the most common ocular manifestation. Other common ocular presentations included microphthalmos, iris abnormalities, glaucoma and pigmentary retinopathy.[1,5] Cataract, microphthalmos and iris hypoplasia is usually a common combination noticed in different studies.[6] While cataract has been appreciated as a more sensitive ocular sign for CRS, iris hypoplasia is regarded as a more specific ocular sign for CRS.[6]

Iris hypoplasia has been reported many times in association with CRS. It was first described in 1941 and there have been many case series around the world reporting it in CRS patients.[7] According to the American Academy of Ophthalmology, iris hypoplasia is the term for an iris that has either been eroded or has been prevented from developing properly.[8] In CRS, the pathogenesis is incomplete development. Glaucoma can occur as an early manifestation of CRS, but has also been reported as a complication of long-standing iris hypoplasia.[7] This is one example of how iris hypoplasia can cause ocular problems for the patient.

While data on iris hypoplasia in CRS is present in abundance, data on aniridia in CRS is lacking. In our literature search, we could not find aniridia being reported as an ocular manifestation of CRS in even a single case. Aniridia is defined as the absence of the iris.[9,10] The incidence has been reported between 1 per 64,000 to 1 per 96,000 live births.[10] It can occur in isolation, either sporadically or in autosomal dominant fashion, and can also occur as part of a syndrome.[10] It occurs in syndromes such as the WAGR syndrome (Wilm's Tumour, Aniridia, Genitourinary abnormalities and Retardation) and the Gillespie syndrome however it has not been reported with CRS.[9,10] This case report is the first of its kind to report aniridia in a patient with CRS. However, it is likely that the association between aniridia and CRS is a chance event rather than a cause and effect relationship.

Data has suggested that there is a less likely chance of aniridia presenting alone. Usually other ocular abnormalities such as nystagmus, strabismus and reduced vision are present in conjunction.[10] Similarly, CRS also rarely presents with just one ocular manifestation.[5] Iris hypoplasia and aniridia, with time, go on to cause further ocular problems in patients, with the most common being glaucoma.[7,10] This highlights the importance of picking up iris abnormalities so that further loss of visual acuity can be prevented timely.[7,11]

CONCLUSION

In a nutshell, besides systemic manifestation CRS also has many ocular manifestations. Iris hypoplasia has been reported repetitively in CRS cases around the world. Complete aniridia has not been reported and this is the first such case report. Aniridia can lead to ocular complications such as glaucoma, nystagmus and photophobia. Patients with aniridia need to have regular, life-long and careful eye check-ups aimed at preventing the complications of aniridia and arresting further loss of visual acuity.

Consent: Written informed consent to share information and images in this case report was obtained from the patient's parents prior to publication.

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Submitted: 7 August, 2017	*Revised: 31 October, 2018*	*Accepted: 7 November, 2018*

Address for Correspondence:
Syed Rehan Ali, P.O.Box 3500, Stadium Road, Karachi 74800-Pakistan
Email: drsrali@gmail.com