



## The importance of fundus eye testing in rubella-induced deafness



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### ABSTRACT

**Objective:** The purpose of this study was to establish a new approach to improve detection of deafness due to rubella.

**Methods:** Colombian institutes for the deaf were visited by a medical team to perform in all enrolled individuals an ophthalmological examination with emphasis in fundus eye by a retina specialist. In cases where ocular alterations compatible with CRS were found, a medical interview by a clinical geneticist analyzing pre-and postnatal history and a thorough medical examination was done.

**Results:** A total of 1383 deaf institutionalized individuals were evaluated in 9 Colombian cities in the period of 2005 to 2006, finding a total of 463 positive cases for salt-and-pepper retinopathy (33.5%), in which rubella could be the etiology of deafness. Medellín, Cartagena, Bucaramanga and Barranquilla were the cities with the highest percentage of Congenital rubella, corresponding to 22.8% of analyzed population. The analysis performed on cases in which reliable prenatal history was obtained in a second appointment ( $n = 88$ ) showed association between positive viral symptoms during pregnancy and salt-and-pepper retinopathy in 62.5% of cases, while both (retinopathy and viral symptoms) were absent in 29.5% of cases; showing a correlation in 92% of cases.

**Conclusions:** The frequency of deafness by rubella obtained by this study is significantly high compared with previous Colombian studies and with international reports. It was possible to correlate the antecedent of symptoms during pregnancy with the presence of salt-and-pepper retinopathy in this deaf population when reliable prenatal history was available, therefore eye testing with emphasis in fundus examination is a good indicator of rubella induced deafness. We propose a new approach in the search of deafness causes, based on a thorough ophthalmologic examination in all deaf people.

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## 1. Introduction

Deafness is a common sensorial deficit in the world; one of every 500 to 1000 newborns has permanent bilateral profound hearing loss [1]. During the recent years the frequency of common etiologies of deafness have changed, due to impact of declining prenatal infections, including rubella, while the increase of genetic causes is evident, secondary to the improvement of molecular testing, among others [2].

Congenital rubella syndrome (CRS) presents with a classic triad characterized by congenital cardiopathy, sensorial deafness (uni

or bilateral) and ocular defects. Maternal rubella infection may pass unnoticed, and around 50% of newborns with CRS have no apparent clinical signs of infection. Hearing loss is present in 60% of CRS cases, most frequently is sensorineural, congenital, profound, bilateral and flat, and may be progressive in nature [3–6]. The spectrum of ocular manifestations includes nuclear cataract, microphthalmia, iris atrophy, glaucoma, strabismus, hyperopia (rather than myopia) and salt-and-pepper retinopathy (also described as pigmentary retinopathy). The coexistence of microphthalmia with bilateral cataract has the poorer prognosis [3].

Salt-and-pepper retinopathy is present in 40–60% of CRS cases, is described as a nonprogressive pigmentary retinopathy (rare cases of progression have been reported) with a few changes at fundus examination similar to those described in retinitis pigmentosa. Generally it's benign, does not affect visual acuity and is commonly located in the posterior pole. In a low percentage

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of cases vision may be compromised when neovascularization develops [3,7,8].

In a previous study in 1992, our team searched for causes of deafness in a Colombian population. Data was obtained during a nationwide screening program in institutes for the deaf; 16 institutes in 9 Colombian cities were evaluated. Among 1715 deaf individuals assessed in Colombia until 1992, the estimated frequency obtained of deafness due to rubella, based on the presented of prenatal viral symptoms and ocular manifestations of CRS, was 26% [9]. Later Colombian studies in children with presumed non-syndromic deafness showed a 19.9% of possible rubella-induced deafness [10].

According to the WHO monitoring system in its global summary of 2012, in Colombia 2597 cases of rubella were notified between 1999 and 2011; over the last 4 years only suspect cases have been reported, none confirmed. The last cases of CRS were in 2005, a total of 5 cases [11]. This is a result of increased immunization coverage in Colombia, recent data showed a coverage rate of 95.2% [12].

There has been remarkable progress in the Americas toward reaching rubella eradication and prevention of CRS, but due to the presence of other endemic regions in the world, the region continues to be in danger of importation of virus, like the ones responsible for the outbreaks of 1998–1999 in Argentina, Bolivia and Dominican Republic or in Haiti in 2000–2011 and in Colombia and Venezuela during the outbreak of 2001–2002 [13].

Usually, CRS is considered as a possible diagnosis in newborns with a spectrum of clinical findings compatible with this prenatal infections, and is confirmed by virus detection or serologic testing during the first year of life (period in which the laboratory testing are sensible and specific) [6,14]. Keeping in mind that about 50% of the newborns with CRS will not exhibit clinical sign of infection [3–6], and that in countries such as Colombia, the diagnosis of hearing impairment is frequently made after the first year of life; the possibility of diagnose a newborn with rubella-induced deafness using the traditional approach is low. The aim of this study is to establish a new approach to improve detection of rubella-induced deafness in children older than 1-year-old and previously classified as non-syndromic hearing loss cases.

## 2. Methodology

This is an observational, cross-sectional study, conducted in institutionalized deaf children from 9 Colombian cities, since November 2005 until May 2006, aiming to establish the prevalence of rubella-induced deafness in this population. The range of severity of deafness in the studied population was severe to profound. It was expected since the population was selected

among institutions for the deaf, which include more severely hearing impaired children than other settings, such as regular schools. The project was approved by the ethics committee of *Pontificia Universidad Javeriana* and *Fundación Oftalmológica Nacional*, and an informed consent was obtained in each case.

The screening began with a first contact with institutions for the deaf in 9 Colombian cities, excluding the Capital city (Bogotá). These institutions were found through the “Instituto Nacional Para Sordos” (National Institute for the Deaf) database, in which all Colombian institutions for the deaf are registered. We visited all existing Colombian institutions for the deaf outside Bogotá, except one, which did not consent to this research; for a total of 13 institutions, 9 public and 4 private.

Ophthalmologic evaluation by indirect funduscopy with previous pupil dilation was performed for all deaf students. The exam consisted of external eye examination and slit lamp evaluation of anterior chamber and lens opacities, practiced by expert ophthalmologist specialize in retina. We were looking for specific findings of rubella as pigment epithelium abnormalities (aspect “salt and pepper” confluent pigmentation, mottled black with depigmented patches). In cases in which ocular alterations compatible with CRS were found, a medical interview and a physical examination by a clinical geneticist was conducted to describe each case completely. The interview included the analysis of pre- and postnatal history, family history including a pedigree emphasizing previous cases of deafness or visual impairment in the family, and a thorough medical examination describing any minor or major malformations found. If the assessment reveals a clear pattern of inheritance or was able to classify the child in a syndrome known for the association of deafness and visual impairment (e.g. Refsum disease, Alström syndrome, Bardet–Biedl syndrome, among others) the case was excluded.

A frequency distribution analysis by means of percentages was conducted.

## 3. Results

A total of 1383 institutionalized deaf people, born between 1982 and 2002, were evaluated in 9 Colombian cities since November 2005 until May 2006. A total of 463 individuals had salt-and-pepper retinopathy (Fig. 1) corresponding to a 33.5% (Tables 1 and 2). Analyzing the prevalence of rubella per city, a distinctly higher percentage of CRS was found in Medellín, Cartagena, Bucaramanga and Barranquilla, accounting for 22.8% of the analyzed population and for 68.2% of rubella cases (Table 3). Of those 463 cases, 2.57% had additional findings of CRS; 1.51% had salt-and-pepper retinopathy and cataract; 0.63% had

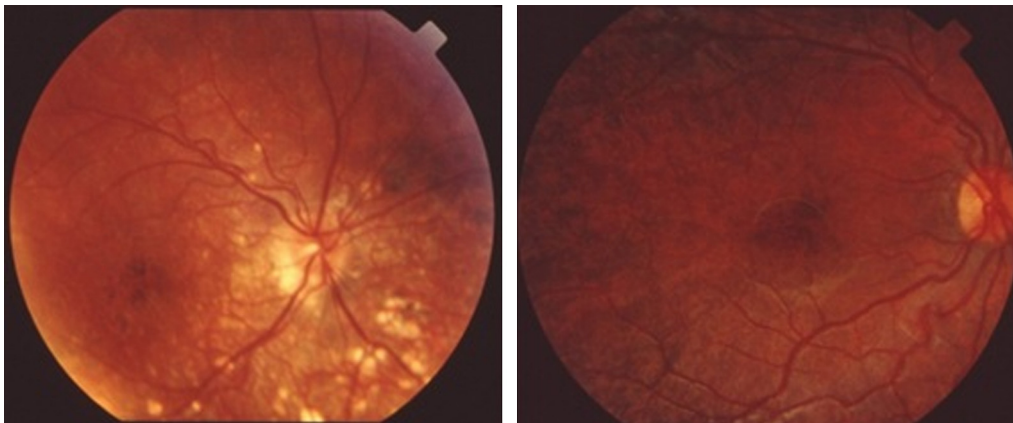


Fig. 1. Salt-and-pepper retinopathy secondary to rubella virus.

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