



## A CASE REPORT ON A RARE ANOMALY: BARTSOCAS PAPAS SYNDROME

RUGMINI KAMMALAMMAL<sup>1</sup> AND SURESH KIRUPANANDHAN<sup>2\*</sup>

*Department of Paediatrics, Sree Balaji Medical College & Hospital (Bharath University) Chennai, India.*

### ABSTRACT

Bartscoas Papas syndrome (BPS) presents a rare and lethal anomaly characterised by severe popliteal pterygium, mid facial defects, syndactyly and genital abnormalities. The purpose of presenting this case is its rarity and its presentation with normal amount of liquor and normal foetal movements which is not hitherto reported. Previous reported cases which were diagnosed during the antenatal period had family history of similar disorder. But this case had no history of similar anomaly in the family.

**KEYWORDS:** Pterygium, Bartscoas Papas Syndrome



**SURESH KIRUPANANDHAN**

Department of Paediatrics, Sree Balaji Medical College &  
Hospital (Bharath University) Chennai, India.

*\*Corresponding author*

## INTRODUCTION

Bartsocas Papas syndrome (BPS) is an Autosomal recessive disorder<sup>1</sup> (OMIM Number 263650). Among the different types of syndromes presenting with pterygiums this form is severe and lethal. It is characterized by marked popliteal pterygium associated with multiple congenital malformations like orofacial clefts and syndactyly. Recently the gene for this malformation was mapped at chromosomal region 21q22.3<sup>2</sup>. Majority of these cases die in-utero or shortly after birth<sup>3</sup> with exceptions reported recently<sup>4,5</sup>. This syndrome has been earlier reported in various parts of world like Bedoin community in Qatar<sup>3</sup>, in Egypt<sup>4</sup>, Mahadeo Koli community in India<sup>6</sup>, but this is a first case to be from South India, to the best of our knowledge.

## CASE HISTORY

A 27 year old lady who is a second gravida with 5 months amenorrhoea underwent medical termination of pregnancy in our hospital as an anomaly was diagnosed by antenatal ultrasound. This conception was out of third degree consanguineous marriage. There was no systemic illness or infectious disease during

her pregnancy, there was no exposure to any drugs or radiation. The quickening was felt well at 5<sup>th</sup> month of gestation. Anomaly scan done at 5 months showed a fetus of 20 weeks gestation with no pre-maxillary triangle made out, lips and palate were not well formed, and inter ocular distance was increased. There was a median facial cleft. Fingers and toes were not well imaged with deformity of hands and feet. In addition to this there was left hydronephrosis and right pelvi-ureteric junction obstruction. Liquor was normal in amount. The abortus was a dead foetus of weight 450grams and length 22cms and the placental weight was 100 grams. The foetus had multiple anomalies, like bilateral popliteal pterygium extending from the thigh to the ankle and also pterygium between the upper arm and chest. There was syndactyly of the toes. Hands bilaterally showed oligo syndactyly. Associated anomalies were macrocephaly, hypertelorism, proptosis of left eye, low set ears, midfacial abnormality with malformed nose, fused oral cavity with filliform bands between the jaws, retrognathia, and wide spaced nipples. Genitals were not well differentiated. Anus was patent. Neck and spine were normal. (Figure 1 and 2)



**Figure 1**  
***Showing the facial anomalies in the foetus***  
***(product of a pregnancy terminated by induced abortion).***



**Figure 2**  
***Showing the popliteal pterigyum (product of a pregnancy terminated by induced abortion).***

Radiological examination showed a normal bony structure. Ultrasound scan of the abdomen didn't show any malrotation or diaphragmatic hernia but there was hydronephrosis of the left kidney.

There was no family history of congenital anomalies and the first sibling was normal.

## **DISCUSSION**

The term arthrogyrosis means curved joint<sup>1</sup>. The term pterygium denotes a wing like structure, a web, or triangular membrane formed across a body joint. The multiple pterygium syndromes are best examples of arthrogyrosis involving limb plus other body areas. Pterygial syndromes are classified according to mode of inheritance and anomalies<sup>1</sup>. Further, pterygial syndrome could be inherited as autosomal dominant or autosomal recessive condition; In case of autosomal dominant condition it may present as popliteal pterygium; antecubital pterygium; and lethal multiple pterygium. Whereas, autosomal recessive condition includes, lethal popliteal pterygium (Bartsocas Papas syndrome); multiple pterygia; multiple pterygia, Escobar type and pterygium and malignant hyperthermia. BPS was mentioned in the literature as early as year 1600, but it was well

described by Bartsocas and Papa in 1972<sup>7</sup>, with anomalies characterized by cleft palate, with or without cleft lip, lip pits, popliteal and other webs, toe nail dysplasia, syndactyly, aplasia of the labia majora. Associated with this mental retardation has also been reported<sup>7</sup>. There is also associated oligodactyly, genital anomalies<sup>3</sup> hypoplastic heart and lungs, cystic hygroma, cardiac hypertrophy, congenital cataract, omphalocele and aplasia of the urethra. The facial features includes micrognathia, low set ears, mid facial defect with hypoplastic nose, small mouth, orofacial cleft with filiform bands across the jaws, absent oral cavity, hypertelorism, short palpebral fissure, ankyloblepharon, and hypoplastic nose<sup>3</sup>. Antenatally, it usually presents with absence of foetal movements and severe polyhydramnios but according to a report<sup>6</sup> oligohydramnios was also a presentation in three consecutive pregnancies in a family. Karyotyping is usually normal in these cases. Our case had all the above features of Bartsocas Papas syndrome with multiple pterygium, with anomalies of face, hands, feet, genitals and skin. There were two exceptional features in our case; normal foetal movements were felt at 5 months of gestation and normal amount of liquor. The pathophysiology of this syndrome is not well understood even though an abnormal fragile collagen is postulated to be

the reason for the early foetal akinesia leading to improper development of the foetal skin and multiple pterygium<sup>6</sup>. The skeletal muscle bulk has been shown reduced with a remaining muscle vacuolar degeneration, dystrophy, a generalized or patchy myotubular appearance, and generalized hypotrophy<sup>8</sup>. Genetic studies in the past have revealed various genes involved in this disorder. Direct sequencing of *RIPK4* (receptor-interacting serine/threonine kinase protein 4) showed a homozygous transversion (c.362T>A) that causes substitution of a conserved isoleucine with asparagine at amino acid position 121 (p.Ile121Asn) in the serine/threonine kinase domain of the protein.<sup>2</sup> Germline-inactivating mutations in the embryonal acetylcholine receptor  $\gamma$  subunit (*CHRNA3*) in families with both lethal and nonlethal forms of the disease has been

reported<sup>9</sup>. Interferon regulatory factor 6 gene mutations have also been reported as a cause of this anomaly<sup>6</sup>. Serial ultrasonography (USG) beginning early in the pregnancy is advocated. The prenatal diagnosis of this syndrome is suspected when two-dimensional ultrasound (2DUS) scan shows several malformations. The three-dimensional ultrasound (3DUS) in rendering mode permits the spatial visualization of these malformations, allowing better understanding of this anomaly by parents. The first prenatal diagnosis of BPS in the first trimester was reported by Dolan *et al*<sup>5</sup>. Further, increased nuchal translucency; absence of limb movements; multiple joint contractures, and cutaneous webs have allowed the diagnosis of lethal multiple pterygium syndromes in the first trimester of pregnancy<sup>10</sup> and cystic hygroma in the second and third trimesters.

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