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Bartsocas-Papas syndrome: The first case report of severe autosomal recessive form from Indonesia

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ABSTRACT

INTRODUCTION: Bartsocas-Papas syndrome (BPS) is an autosomal recessive form of Popliteal Pterygium syndrome (PPS). It is a very rare disease characterized by congenital craniofacial anomalies, popliteal webbing, and genitourinary and musculoskeletal anomalies. Almost all of the cases were reported in dead intrauterine pregnancies.
PRESENTATION OF CASE: We present a 10-month-old boy with bilateral complete cleft lip and palate, abnormal scalp hair, an absence of both upper eyelids, choanal atresia, syndactyly of the third and fourth fingers of the right hand, agenesis fingers on the left hand, bilateral popliteal pterygia, bilateral talipes equinovarus, agenesis of the toes of both lower extremities, intercrural webbing, an absence of testis, and scrotal anomaly. Multistage surgical correction was performed for the multiple congenital malformations.
CONCLUSION: We report the first case of BPS from Indonesia. Gradual management should be performed according to the patient's age and available facilities.

1. Introduction

Bartsocas-Papas syndrome (BPS) is a very rare congenital syndrome with autosomal recessive inheritance, variable expressivity, and incomplete penetrance. The first case was reported by Trelat in 1869 [1]. The three main clinical manifestations are craniofacial malformations (orofacial cleft, short palpebra fissure, ankyloblepharon, hipoplastic nose), muscle-skeletal anomalies (bilateral intracrural popliteal pterygia, various digital deformities), and genitourinary anomalies (cryptorchidisme, agenesis testis or cleft scrotum, inguinal hernia) [2]. Almost all of the cases reported were dead neonates or dead intrauterine pregnancies [3]. We report the first case of BPS case from Indonesia according to the 2020 Surgical Case Report guidelines [4].

2. Case report

A 10-month-old boy was referred to our institution from Borneo, Indonesia, with a diagnosis of BPS. A traditional midwife helped with the normal delivery because the family lived in the middle of the forestland of Borneo out of the reach of medical facilities. His weight was 2200 g at birth. He was the second child of non-consanguineous parents. There was no history of medication taken during pregnancy and no history of the congenital anomalies observed. His mother, brother, and maternal grandfather have lower lip pits. As noted on the patient's pedigree (Fig. 1), the genetic pattern showed that his maternal grandfather, mother, and brother had two pits on their lower lips.

The patient had multiple congenital anomalies, such as clefts of the soft palate, hard palate, and lips; abnormal scalp hair (hirsutism) in the frontal area; ectropion of both upper eyelids; and bilateral choanal atresia (Fig. 2A). Examination of the upper extremities exhibited syndactyly of the second and third fingers, reduction defect of the thumb, agenesis of the fifth digit of the right hand, and only two fingers on his left hand.

The lower extremities showed bilateral popliteal pterygia resulting in tenting of the soft tissue with incomplete extension at the knees, as well as bilateral talipes equinovarus and agenesis of the digits of both lower extremities (Fig. 2B). Intercrural webbing

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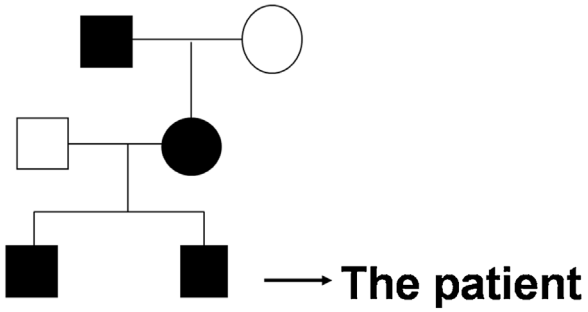

Fig. 1. Pedigree of the patient's family.

was noted from the medial thighs to the base of the penis, giving the scrotum a diminutive and bifid appearance (Fig. 3). The testes were absent, and the penis was 4 cm in length with the external urethra orifice in its normal position.

Laboratory findings were normal. Urology ultrasonography was unable to reveal the testes appearance clearly, but the other internal organs were normal. Roentgenographic examination of the upper and lower extremities showed syndactyly; reduced numbers of carpal, metacarpal, and phalange bones; and fusion of the metatarsals (Fig. 4A). There were no cardiac anomalies. Ophthalmological examination revealed a cornea descemetocele in the right eye and exposure keratitis in the left eye. A chromosomal study demonstrated chromosome 14/5 translocation t (14; 5) (5p) and gap 6p non-specific (Fig. 4B).

Our team of plastic and reconstructive surgeons performed bilateral ectropion release and a full-thickness skin graft in the first stage of reconstruction (Fig. 5A). Gastrostomy was performed by pediatric surgeons due to nutritional problems. In the second stage of reconstruction, we inserted three tissue expanders in both cheeks and the forehead and maintained them for three weeks. In the third stage, we performed soft-tissue facial reconstruction, and then his cleft palate was closed at 12 months of age (Fig. 5B).

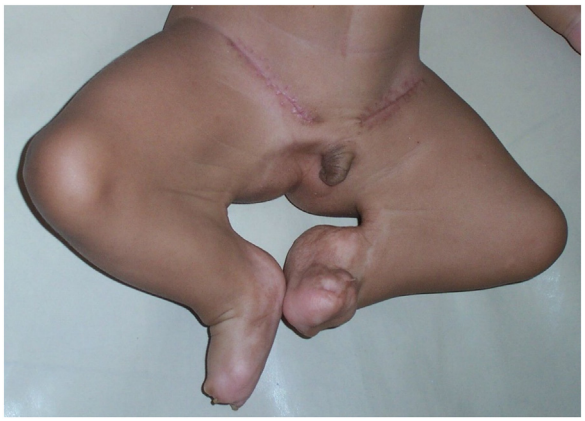

Fig. 3. Popliteal webbing bilateral and intercrural webbing.

Post-surgery, broad-spectrum antibiotics and analgesic drugs were administered for five days. The patient was discharged from the hospital in an optimal condition. Further reconstruction entailing soft tissue release of the knees will be performed at 24 months of age.

3. Discussion

BPS is a syndrome characterized by bilateral popliteal webbing and affects three anatomical areas: the facial, genital, and popliteal areas [3]. Hale classified pterygium syndromes into autosomal dominant and recessive form, of which the latter includes two lethal forms of PPS: BPS and multiple pterygium syndrome [5]. Even though the etiology is unknown, this syndrome is related to an autosomal dominant or recessive gene mutation with incomplete penetrance and variable expressivity [6–8]. There are widely variable degrees of manifestations with which those carrying the mutation may be affected, even within families, as shown here.

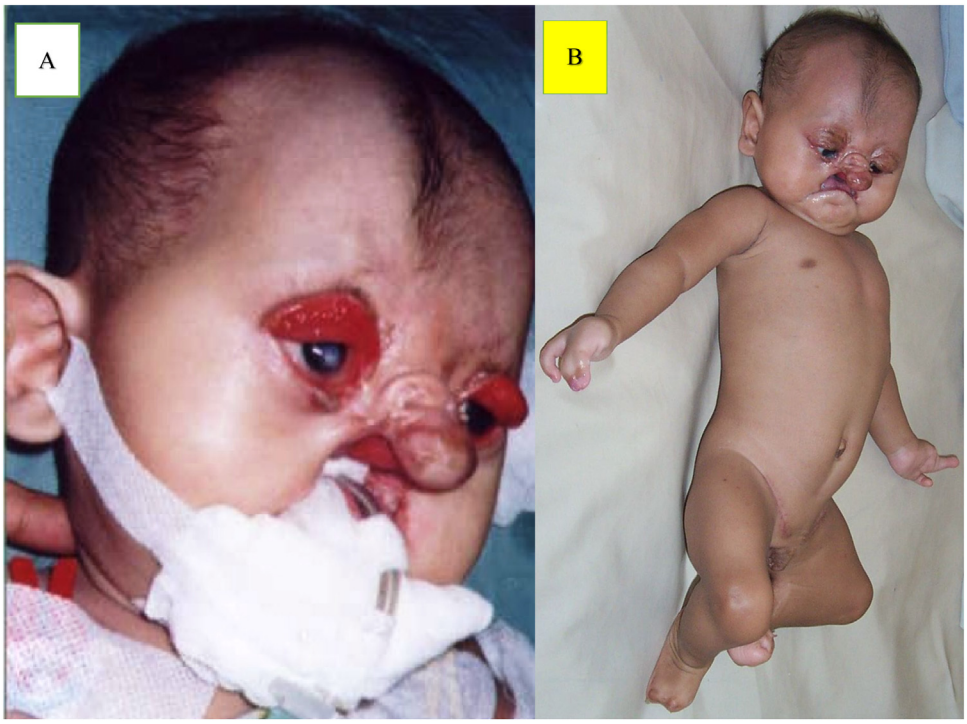

Fig. 2. A) Cleft lip, cleft palate, hirsutism, atresia of both upper eyelids, short nose, choanal atresia. B) Bilateral talipes, agenesis toes of both lower extremities, agenesis of testes, scrotal anomaly, and digital anomalies on the right hand.

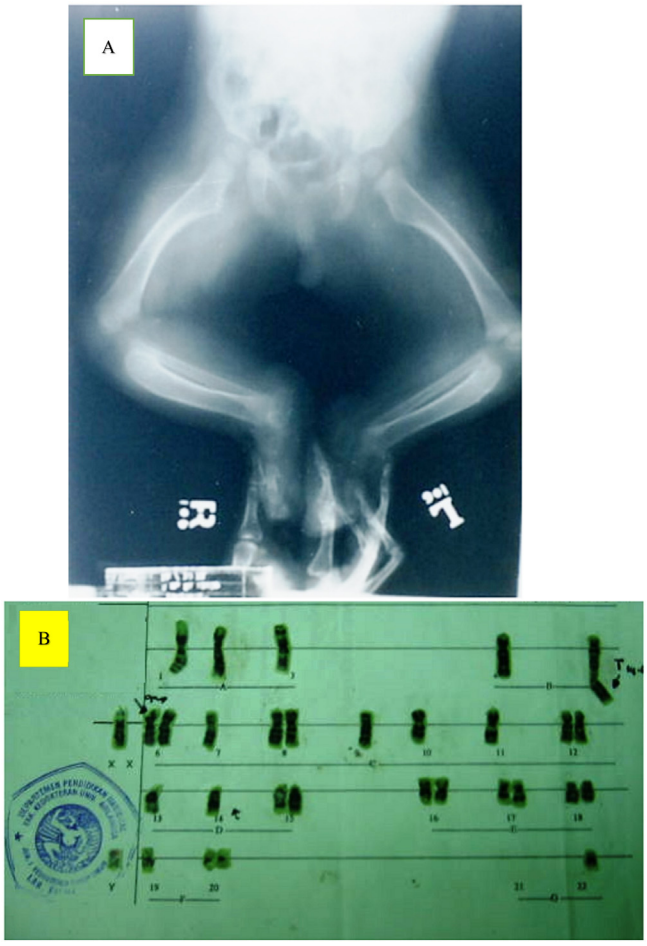

Fig. 4. A) Roentgenographic examination of the lower extremities. B) Chromosomal analysis: 46 XY, t (14;5) (5p) gap 6p non-specific.

Therefore, careful clinical examination of parents and relatives may be necessary.

The clinical manifestations of include craniofacial anomalies such as abnormal scalp hair, atresia of the external ear canal, cleft lip and palate (90%), salivary lower lip pits (46%), an intraoral fibrous band connecting the maxillary and mandible alveolar ridge (43%), oral frenula, hypodontia, and cutaneous webs between the eyelids (20%). Musculoskeletal anomalies include popliteal webbing (90%), toe displasia (30%), syndactyly of the toes and fingers, intercrural pterygium (9%), hypoplasia or aplasia of the digits, reduction defect of the thumb, fusion of distal interphalangeal joints, valgus deformity of the feet, hypoplasia of the tibia, bifid or absent patella, posterior dislocation of the fibulae, spina bifida occulta, other vertebral anomalies, bifid ribs, short sternum, and scoliosis. Genital anomalies include hypo plastic labia majora, scrotal dysplasia, cryptorchidism, ambiguous external genitalia, penile ectopia or torsion, ectopic testes, underdevelopment of the vagina or uterus, and inguinal hernia. There is no disturbance in growth or intelligence [3,6,7].

Differential diagnosis between BPS, PPS, and Van der Woude syndrome (VWS) is sometimes very difficult [9]. Features of VWS have been seen in individuals with PPS or BPS and have also been linked to chromosome band 1q32. These three entities may represent a contiguous gene deletion syndrome or allelic variants. VWS typically consists of a cleft lip or palate and distinctive pits of the lower pits. Extra-oral manifestations are rare, but limb anomalies, popliteal webs, brain abnormalities, congenital heart defects, and Hirschsprung disease have been reported [7,9]. Management of BPS

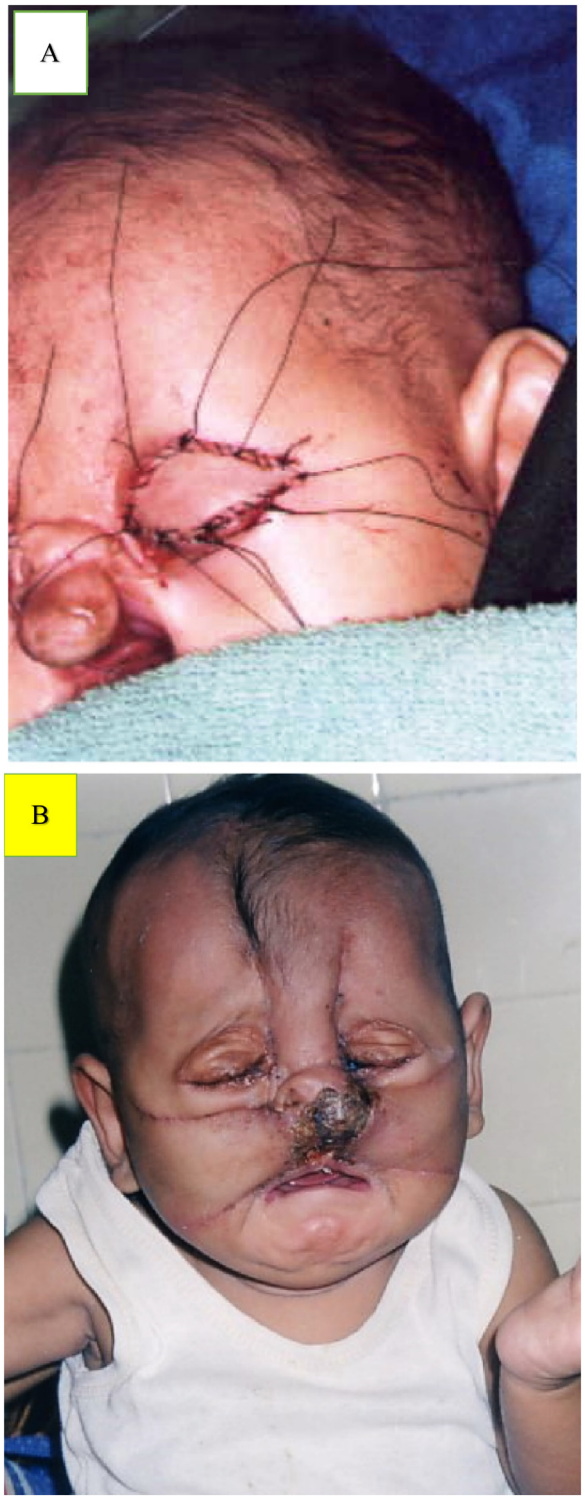

Fig. 5. A) Release ectropion and full-thickness skin graft. B) After three stages of facial soft tissue reconstruction.

is best performed by a multidisciplinary team of plastic surgeons, pediatric surgeons, pediatric urologists, anesthetists, pediatrics, otolaryngologist, geneticists, dentists, physical therapists, social workers, and speech therapists.

4. Conclusion

We have reported the first case of BPS from Indonesia. Almost all of the cases reported have involved dead intrauterine pregnancies. Gradual management according to the patient's age and available facilities should be performed. BPS should be handled by a multidisciplinary team for optimal management.

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Ethical approval

The study is exempt from ethical approval in our institution.

Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Author contribution

Fonny Josh, Tomie H. Soekamto, and Djohansjah Marzoeki: study concept, surgical therapy for this patient. Fonny Josh, Tomie H. Soekamto, and Djohansjah Marzoeki: Data collection, Writing-Original draft preparation. Djohansjah Marzoeki: senior author and the manuscript reviewer. Muhammad Faruk and Fonny Josh: Editing, Writing. All authors read and approved the final manuscript.

Registration of research studies

Not applicable.

Guarantor

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Declaration of Competing Interest

The authors report no declarations of interest.

References

[1] C.S. Bartsocas, C.V. Papas, Popliteal pterygium syndrome. Evidence for a severe autosomal recessive form, J. Med. Genet. 9 (1972) 222–226, http://dx.doi.org/10.1136/jmg.9.2.222.
[2] Indian Pediatrics - Editorial Bartsocas papas syndrome, (n.d.).
[3] E.M. Abdalla, H. Morsy, Bartsocas-Papas syndrome: unusual findings in the first reported egyptian family, Case Rep. Genet. 2011 (2011) 1–6, http://dx.doi.org/10.1155/2011/428714.
[4] R.A. Agha, T. Franchi, C. Sohrabi, G. Mathew, A. Kerwan, A. Thoma, A.J. Beamish, A. Noureldin, A. Rao, B. Vasudevan, B. Challacombe, B. Perakath, B. Kirshtein, B. Ekser, C.S. Pramesh, D.M. Laskin, D. Machado-Aranda, D. Miguel, D. Pagano, F.H. Millham, G. Roy, H. Kadioglu, I.J. Nixon, I. Mukhejree, J.A. McCaul, J. Chi-Yong Ngu, J. Albrecht, J.G. Rivas, K. Raveendran, L. Derbyshire, M.H. Ather, M.A. Thorat, M. Valmasoni, M. Bashashati, M. Chalkoo, N.Z. Teo, N. Raison, O.J. Muensterer, P.J. Bradley, P. Goel, P.S. Pai, R.Y. Afifi, R.D. Rosin, R. Coppola, R. Klappenbach, R. Wynn, R.L. De Wilde, S. Surani, S. Giordano, S. Massarut, S.G. Raja, S. Basu, S.A. Enam, T.G. Manning, T. Cross, V.K. Karanth, V. Kasivisvanathan, Z. Mei, The SCARE 2020 guideline: updating consensus surgical CAse REport (SCARE) guidelines, Int. J. Surg. 84 (2020) 226–230, http://dx.doi.org/10.1016/j.ijsu.2020.10.034.
[5] E. Kalay, O. Sezgin, V. Chellappa, M. Mutlu, H. Morsy, H. Kayserili, E. Kreiger, A. Cansu, B. Toraman, E.M. Abdalla, Y. Aslan, S. Pillai, N.A. Akarsu, Mutations in RIPK4 cause the autosomal-recessive form of popliteal pterygium syndrome, Am. J. Hum. Genet. 90 (2012) 76–85, http://dx.doi.org/10.1016/j.ajhg.2011.11.014.
[6] E.J. Leslie, J. O'Sullivan, M.L. Cunningham, A. Singh, S.L. Goudy, F. Ababneh, L. Alsubaie, G.-S. Ch'ng, I.M.B.H. van der Laar, A.J.M. Hoogeboom, M. Dunnwald, S. Kapoor, P. Jiramongkolchai, J. Standley, J.R. Manak, J.C. Murray, M.J. Dixon, Expanding the genetic and phenotypic spectrum of popliteal pterygium disorders, Am. J. Med. Genet. Part A 167 (2015) 545–552, http://dx.doi.org/10.1002/ajmg.a.36896.
[7] E.-C. Tan, E.C. Lim, S.-T. Lee, De novo 2.3 Mb microdeletion of 1q32.2 involving the Van der Woude Syndrome locus, Mol. Cytogenet. 6 (2013) 31, http://dx.doi.org/10.1186/1755-8166-6-31.
[8] J.M. Graham Jr, P.A. Sanchez-Lara, Smith's Recognizable Patterns of Human Deformation, 4th ed., Elsevier Inc, 2016.
[9] R.D. Bennun, E. Stefano, L.E. Moggi, Van der Woude and Popliteal Pterygium Syndromes, J. Craniofac. Surg. 29 (2018) 1434–1436, http://dx.doi.org/10.1097/SCS.0000000000004698.