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Research Article

A Multidisciplinary Approach To Poland Syndrome: Experiences From A Novel Clinical Approach.

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Abstract

Aims: Poland syndrome is a rare but complex multifactorial condition related to the development of the upper limb bud. It can present with differences of the hand, chest wall, shoulder girdle, spine and breast tissue. It is also associated with cardiac and renal anomalies. Whilst individual concerns are usually addressed in isolation by separate clinicians, our model of a multidisciplinary clinic is the first in the UK to provide a unified assessment with upper limb, breast and thoracic surgeons reviewing each difference in turn. We aim to describe and discuss the potential benefits of this new service.

Methods: A retrospective review was performed of 42 consecutive patients between February 2022-December 2023, and questionnaires sent evaluating patient experiences.

Results: This demonstrated a mean age at referral of 6 years. Ten were referred from outside the normal catchment area, for which the mean distance travelled was 121 miles. Five had previously undergone surgery for conditions related to Poland Syndrome elsewhere. A survey identified 88.9% felt the clinic setup was beneficial, and 55% felt more knowledgeable about the condition after the appointment. Seventy-seven percent of respondents enjoyed meeting others with the condition.

Conclusion: The aim of the clinic is not to dictate treatment plans, or to dismiss those of others. It aims to allow families to be fully informed about all options, offer support and a presence when patients feel our skills are required. Our experiences and results demonstrate that for rarer conditions such as this, this model gives time and space for families to understand more about the condition, offering a holistic care model empowering families to make the decision that is most suitable for their circumstances.

INTRODUCTION

Poland syndrome (PS) is a complex congenital condition describing a spectrum of differences to the developing upper limb bud. First described in 1841, Alfred Poland remarked upon the absence of pectoral muscles **[1]**, and although typically involving the ipsilateral sternocostal head of the pectoralis major and upper limb, clinical manifestations are variable and can also involve differences to breast tissue, spine, shoulder girdle, thoracic skeleton and musculature. It affects between 1 in 7000 and 1 in 100,000 live births and is two to three times more common in males **[2]**. It predominantly affects the right side **[3]**.

Bavinck and Weaver first described the subclavian artery supply disruption sequence to include Poland syndrome, Klippel-Feil, Sprengel, and Möbius anomalies **[4]**. Obstruction of the subclavian artery proximal to the origin of the internal thoracic artery but distal to the origin of the vertebral artery could cause a combination of hand and chest wall defects. This hypothesis is supported by vascular studies, which demonstrated an ipsilateral reduction in subclavian artery diameter and flow velocity **[5]**.

The exact causation of this vascular interruption is unclear and could be caused by intrinsic thrombus or emboli, or extrinsic compression from amniotic band or aberrant musculature **[6]**. Maternal factors such as smoking and cocaine abuse have also been implicated **[7, 8]**. Finally genetic mutations in thrombophilia genes **[9]**, or those related to cellular growth and apoptosis have been postulated as causative **[10]**.

Although some cases are identified during neonatal checks, due to the rarity of the condition and often mild phenotype, the condition may go unrecognised into puberty where breast hypoplasia can be of concern. Baldelli et al reported a mean age at diagnosis of 14 years **[11]**.

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Due to the wide variety of psychological, functional and cosmetic concerns from the affected families, and subspecialisation of surgical expertise, patients can be referred to several different clinics, meaning numerous appointments which patients and guardians may find overwhelming. Feedback on specialist centres for rare conditions is positive, reducing the number of appointments and avoiding the need to repeatedly explain their condition **[12]**.

We have developed the first UK multidisciplinary model, in which thoracic, breast and hand surgeons review the patient together and provide a holistic approach considering not only their physical health but also their emotional, social, and psychological well-being. We present our experience in building a successful and reproducible multidisciplinary PS program as well as

present our model and feedback from patients/guardians reported outcomes who

attended our new clinic.

The Clinic

The clinic, run at a Regional Specialist Children's Hospital, accepted referrals from nationwide sources including General Practice, Plastic Surgery colleagues, and Paediatricians.

Patients with parents are given a 1-hour time slot. The consultation and examination are carried out by a Paediatric thoracic surgeon, a Plastic and breast reconstructive surgeon and a surgeon specialising in congenital upper limb and hands anomalies. An upper limb physiotherapist is also present, as is a representative from PIP-UK, a Poland syndrome support group **[13]**. PIP-UK is a non-profit, registered charity which advocates for earlier and structured diagnostic and therapeutic pathways for PS patients, building awareness and providing support for the PS community

A plain chest x-ray to rule out dextrocardia, and renal ultrasound to exclude renal anomalies are usually pre-arranged, to be performed on the day of clinic attendance. After the consultation, parents and older children were given the opportunity to spend time with the PIP-UK representative who offered further information on the charity, the PS community and meet other children or adults with PS in a social setting.

Patients were sent a follow up feedback questionnaire (Appendix A) within 2 weeks of the appointment, distributed via AccurX (AccurX, London, UK), which was approved by all stakeholders.

RESULTS

Forty-one patients were reviewed between February 2022-December 2023. Patient demographics are presented in **Table 1**. One case was misdiagnosed on referral, and following review in clinic was found to have a fully functional and intact pectoralis muscle and was therefore excluded. Two patients did not attend their clinic appointment. Fifteen of the 38 patients attending the clinic were female (39.4%), with a mean age of 7.0 years and median of 7.0 years. The left side was affected in 19 patients (50%).

The mean distance travelled for all patients was 75.4 miles, however twenty patients were referred from outside the normal catchment area for the hospital and would routinely be referred to a local unit for management. The mean distance travelled for these patients was 121.2 miles (**Figure 1**). At time of writing, we have also received enquiries and request for a consultation from a family based in the Republic of Ireland. This child and his family had become aware of this multidisciplinary clinic thanks to the PIP-UK network.

Table 1. Demographic details of patients attending the clinic.

Age at referra($\bar{x}$)	6.9
Range	0-15
Sex	
M	23
F	15
Source of referral	
GP	26
Paediatrician	7
Obstetrician/Neonatologist	2
Local Plastic Surgeon	3
Mean distance travelled (miles) out of region	129.75 (53-257)
Mean of all	75.44 (1.8-257)
Side affected	
Right	19
Left	19
Previous surgery for Poland issue	
Y	7
N	31
Chest (n)	
No difference	1
Pectoralis Major hypoplasia	9
Pectoralis Major aplasia	28
Nipple Absence/Asymmetry	1
Latissimus dorsi hypoplasia/aplasia	2
Dextrocardia	1
Pectus Carinatum/Excavatum	7
Rib hypoplasia/aplasia	2
Breast tissue asymmetry	4
Spine (n)	
No Difference	34
Mild Scoliosis	4
Renal (n)	
No Difference	22
Hydronephrosis	3

Figure 1. Geographical location of patients referred to the Poland Syndrome clinic at Birmingham Children's Hospital (BCH). The location of BCH is annotated with a yellow circle. (Made with Google My Maps, Google, California, USA) Map data ©2024 GeoBasis-DE/BKG (©2009), Google.



Thirty-six patients (94.7%) had absence or hypoplasia of the pectoralis major muscle, one patient had a hypoplastic latissimus dorsi (LD), and one had dextrocardia. One patient had hypoplastic breast tissue with mild hypoplasia of the hand and forearm, and one had a subtle ipsilateral ribcage deformity with symbrachydactyly. Seven (18.4%) had sternal anomalies, 3 pectus excavatum, and 4 pectus carinatum. Three patients had mild scoliosis identified clinically (Table 1).

Hand differences were observed in 20/38 (52.6%) patients and assessed using the classification system proposed by Perez-Lopez **[14]** (**Table 2**).

Three of 25 (12%) had minor asymptomatic hydronephrosis diagnosed on renal ultrasound, which was managed with ongoing observation. Thirteen patients were referred locally for investigation and results were not available.

One patient had Poland-Mobius syndrome, with the addition of an Abducens nerve palsy, a much rarer combination with estimated incidence of 1 in 500,000. **[15, 16]**

Table 2. Upper limb differences demonstrated as per Perez-Lopez classification 2022.

Classification	Description	N (%)
Type 1	Isolated pectoralis major aplasia – no upper limb deformity	17 (44.7)
Type 2	Hypoplastic hand without syndactyly	11 (28.9)
Type 3	Hypoplastic hand with syndactyly	10 (26.3)
Type 4	Complete syndactyly including first web space +/- symphalangism	0
Type 5	Syndactyly with longitudinal deficiency of digits/metacarpals	0
Type 6	Transverse amputation at metacarpal levels, affecting ≥1 ray to complete aplasia of the hand	0

Five patients (13.2%) had previously undergone surgery for differences related to Poland's syndrome. Three patients underwent syndactyly release, one had undergone a z-plasty release to the anterior axillary fold, and one patient underwent both syndactyly release and latissimus dorsi transfer to the anterior chest as a toddler. At the time of consultation with us, the flap showed evidence of atrophy.

Seven patients (18.4%) had been signposted towards the clinic from PIP-UK, and two had been seen by specialist paediatric hospitals but were requesting a second opinion.

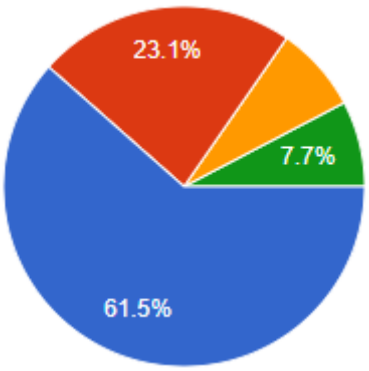
All patients from outside the catchment area were referred locally for ongoing follow up and to arrange basic radiological investigations if they could not be performed during the clinic attendance. Two patients with mild hand differences were given patient-initiated follow-up. Ten further patients were kept for ongoing review.

Survey responses

There were thirteen of 38 responses (34.2%). All thirteen (100%) respondents either agreed or strongly agreed that discussing all aspects of care in one appointment was useful, and all thirteen preferred seeing multiple specialists in a single appointment. Nine responders (69.2%) strongly agreed that they received clear information and fully understood the information that was provided, whereas the remaining four responders were still in agreement.

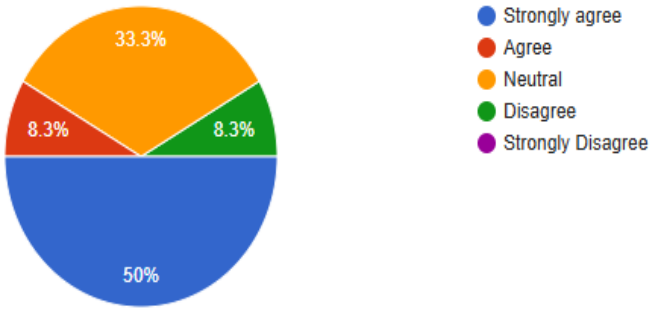
Eight (61.5%) responders strongly agreed that they feel more knowledgeable about the condition following the appointment, and eight felt better positioned to answer their child's questions regarding the condition. One felt less confident (**Figure 2**).

Figure 2. Following the appointment, I have felt better informed to answer my childs' questions about their condition. Total 12 responses.



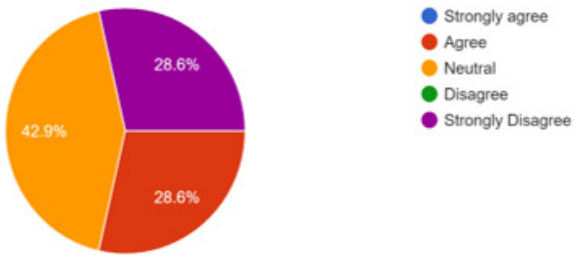
Seventy-seven percent of responders enjoyed meeting others in the waiting area with the same condition, and 50% strongly agreed that their child was more confident after meeting others (**Figure 3**). Eight felt less isolated following the appointment.

Figure 3. My child is more confident as a result of meeting others with the same condition. 12 Responses



Eight responders felt they had adequate time to answer questions. Seven (53.8%) had previously been seen by a specialist regarding their condition. Of these, two (28.5%) responders felt they were now more confused due to the treatment options presented (**Figure 4**).

Figure 4. Following previous consultations, I now feel more confused about how best to manage the condition as there has been more than one option presented. Total 7 responses



DISCUSSION

Due to the nature of the condition, Poland syndrome can involve multiple hospital consultations and investigations to manage the entire spectrum of disorders. A 2015 survey highlighted that patients with rare diseases receive at least one incorrect diagnosis before a final diagnosis is made and are left to research their own condition, often identifying worrying information that negatively impacts mental health. **[17, 18]**. Indeed delay to diagnosis is common **[19]**, and lack of information can lead to frustration, anxiety, low-mood and suicidal thoughts **[20, 21]**. Reproductive planning is also a source of anxiety for families, and as PS is generally considered a sporadic condition, families should be reassured

of the chances of recurrence in a future pregnancy. **[22]** The development of this clinic aimed to reduce the burden on families and improve the patient experience, and the addition of a genetic counsellor and psychologist is a potential area for development.

The development of the multidisciplinary clinic

The function of the multidisciplinary team (MDT) is to bring together clinicians from different disciplines to formulate a management plan for patients. It is commonly employed in healthcare settings, primarily in oncology, serving to improve communication and decision making, sharing information, and consistency in the standard of care offered. **[23, 24]**, however may be beneficial in rare diseases to improve communication, to address the physical and psychological aspects of a condition **[25]**. Very few studies discuss the involvement of the patient or family within these discussions however this seems a valuable progression of the model as it can address the patients' concerns directly rather than feeding back expert opinion and decisions.

The creation of the team relies on understanding the needs of patients and families affected, and the potential resources available to the service. The first step is to create a business plan which involves identifying key stakeholders, defining objectives, and developing a vision for the clinic. It serves to inform the administrative team of the hospital the status quo, how the service can improve the patient journey, and anticipated financial costs.

Following this an engaged team should be assembled including a variety of professionals. In the case of the PS clinic, this included surgeons who are specialists in the most common complaints and concerns, chest wall, upper limb, and breast anomalies. A hand therapist and PIP-UK representative were then recruited, the latter helping with marketing and publicising the service.

The appointment of a coordinator can help with communication and ensures a consistent point of contact for both internal and external referrals.

A framework should be developed to establish responsibilities and contributions of each discipline. Classifications and scoring systems can be useful to standardise treatment and guide decision making, as well as understand more about each aspect of the decision-making process. Several exist for each anatomical area of concern, and the choice should consider ease of use, and how it affects treatment choice. A national consensus survey was performed in Italy to define best practices in the absence of published high-quality evidence for decision making **[26]**.

Key performance indicators can be defined to assist in evaluating impact. Regular feedback from patients and MDT members can aide in identifying areas for development and improvement, which can be a cyclical process. This can involve adjustments in clinic personnel, appointment timings, information provision and logistical changes.

The role of the surgeon

Patients commonly presented at one of three stages. Firstly, in the neonatal period where a diagnosis has been made, and parents seek more information around the condition which may lead to preparing for hand surgery, usually performed around 2 years of age. The second is around 5-7 years old, where patients start to notice a difference between themselves and peers, and parents and patients seek support and reassurance regarding future function and appearance. Lastly the peripubertal child, where hormonal changes and development can affect self-image, and information about options is provided.

The role of the surgeon should not be simply to offer a surgical procedure. In complex and rare conditions, the surgeon should aim to take a holistic approach, offering support, discussing options and directing the patient towards another specialist if this is required. Patients and parents are unlikely to have met another family with the same difference and therefore are unable to obtain the same level of support that may be available to those with a more common condition.

One key benefit of this combined clinic was that patients and parents could meet others with the same condition in the waiting room, which is not normally possible for a general hand or breast outpatient clinic. Key to this was the PIP-UK support worker, who helped introduce families and provide a link to others in similar positions. Seven patient referral letters stated parents had requested referral to this specific clinic, which is most likely due to the circulation and promotion of the clinic from the charity itself, following parents online searching for support and further information. Experience from Italy suggests following the creation of a national Poland syndrome association **[27]**, publicity has led to a dramatic increase in referrals, which may be the case as PIP-UK becomes more established.

Nevertheless, surgical management is often requested and sought. Certain decisions can be made on behalf of the patient, and some delayed until the patient can make an informed decision regarding their own body and priorities. The role of the surgeon is to discuss the risks, benefits, and provide guidance to empower the family to be confident in making the right choice. Choices and priorities can change over time due to changes in personal circumstances and how the patient views themselves. Boys and girls may have different priorities, certainly related to breast development, and each should have an individualised treatment plan. This holistic approach at an early age starts the journey, and patient-initiated follow-up allows patients to contact when we are needed. This also encourages responsibility in themselves which is also empowering.

Timing of surgical intervention

The timing of reconstruction of the chest is controversial, and practice is variable. Anderl and Kerschbaumer proposed early latissimus dorsi transfer to rebalance musculature and prevent worsening scoliosis or chest wall deformities in two male children under 5 years old. **[28]**. It is also utilised for aesthetic purposes to recreate the anterior axillary fold, however, can often be hypoplastic **[29]**, with donor site morbidity. The consensus of the clinic is to delay any reconstruction of the chest wall until the child can partake in the consent process.

Adolescence is a challenging period, especially considering body image, and breast development can be extremely emotive necessitating physical and psychological support. Patients desire symmetry in size, shape and fell, but also compared to others, and become 'normal' **[30]**. Baldelli demonstrated that breast surgery during teenage years can improve negative body image and quality of life, which cannot be overcome by surgery in adulthood **[31]**.

Noisser et al found mean areolar diameter having a significant correlation to subjective assessment and long-term outcome, rather than breast size or scar appearance, in patients with congenital breast asymmetry **[32]**, operating between the ages of 16-42. This suggests a delay to symmetrise with the contralateral developed breast as important.

The technique utilised for breast reconstruction has traditionally involved expander and implant reconstruction alone or in combination with LD **[33,34]**. Disadvantages of these techniques include risks of capsular contracture, breast implant illness, and BIA-ALCL with implants **[35]**, whilst in patients with raised body mass index (BMI), volume of myocutaneous LD may not be sufficient to symmetrise.

Procedures undertaken

Techniques in microsurgery have developed and autologous tissue is recognised as gold standard in oncoplastic breast reconstruction in terms of texture and longer-term stability of appearance [**36]**. A proposed algorithm by Mahrhofer taking severity and BMI into account includes free tissue transfer for more severe cases, and lipofilling for less severe **[37]**. Whereas lipofilling is a minimally invasive procedure and can be repeated as necessary **[36, 38]**, free tissue transfer is a more significant procedure and therefore delay until patients can fully comprehend the risks, and Tanner stage 5 is reached is warranted. The breast pocket can be developed in preparation for this with an expander, which may also satisfy patients need for a breast contour.

Whilst certain differences are more easily hidden beneath clothing, hands are more visible. There are a wide variety of phenotypes associated with Poland syndrome, and how the hand appears subjectively can be as important as the function. Therefore, it is vital to place scars in positions that are not immediately obvious to observers, or risk the child hide the hand away due to self-consciousness.

Investigation of associated conditions

The clinic also screened for associated conditions such as dextrocardia, disorders of the renal system, and skeletal differences, most commonly scoliosis and pectus excavatum and carinatum. Dextrocardia is associated with left-sided Poland syndrome, is associated with ipsilateral partial agenesis of 2 or more ribs **[39]**. Fraser et al conclude that dextrocardia is a consequence of the vascular disruption sequence, rather than the malposition being a causation **[40]**. Overall incidence in literature ranges from 7-12% **[39, 41]**, but of those with left-sided differences, 28% were affected **[39]**. This was screened for with plain radiograph of the chest. Case reports document a range of renal issues, ranging from agenesis and hypoplasia to undescended testes, more commonly ipsilateral. Assadi concluded that as most patients are asymptomatic, screening should be performed on all patients with Poland syndrome with routine ultrasound, a practice adopted by this clinic **[42]**. Scoliosis in Poland syndrome is uncommon. A large review found 2/113 (1.7%) **[41]** patients had clinically evident curvature on physical examination, confirmed by radiograph, which may be related to the imbalance of musculature in the shoulder girdle **[43]**. Our smaller series identified 7% of patients having scoliosis. There also exists a link between Poland syndrome and various malignancies including Wilms tumour, leukaemia, and lymphoma **[44]**. Our service did not routinely screen for these, except for Wilms which would be detected by renal ultrasound.

CONCLUSION

We believe this is the first such clinic in the UK to offer this multidisciplinary model for Poland syndrome, and patients have sought referral from a wide geographical area for this specialist service. Despite this, the treatment options offered were not novel or unique, and these were all available locally. However, this 'specialist clinic' model had the benefits of time, combined expertise and opinion, and the shared benefit of meeting others and discussing issues in the waiting area beforehand, demonstrated by the positive feedback and comments regarding isolation and confidence. Indeed, one further improvement we hope to make is the addition of a psychologist to help children manage their feelings towards their condition, and the aesthetic appearance of their hand and chest, and would be valuable at each stage of presentation.

The challenges with rare conditions are that it is difficult to reach consensus as to optimal treatment, due to paucity of high-quality evidence. We aimed to provide an honest appraisal of various approaches, with advantages and disadvantages of each, which the survey reports support. There exist no agreed

UK standardised treatment and variation exists in timing of surgery, especially breast reconstruction, which can lead to confusion for patients and parents, leaving them to make their own decisions regarding treatment. There is a need for those treating this condition to co-ordinate the approach to managing these patients, ultimately improving the decision-making tools available for the patient and family.

REFERENCES

1. Poland A. (1841). "Deficiency of the pectoral muscles". Guy's Hospital Reports. Vol VI: 191–193

2. Freire-Maia N, Chautard EA, Opitz JM, Freire-Maia A, Quelce-Salgado A. The Poland syndrome-clinical and genealogical data, dermatoglyphic analysis, and incidence. Hum Hered. 1973;23(2):97-104. doi: 10.1159/000152560. PMID: 4356989.

3. Fokin AA, Robicsek F. Poland's syndrome revisited. Ann Thorac Surg. 2002 Dec;74(6):2218-25. doi: 10.1016/s0003-4975(02)04161-9. PMID: 12643435.

4. Bavinck JN, Weaver DD. Subclavian artery supply disruption sequence: hypothesis of a vascular etiology for Poland, Klippel-Feil, and Möbius anomalies. Am J Med Genet. 1986 Apr;23(4):903-18. doi: 10.1002/ajmg.1320230405. PMID: 3008556.

5. Merlob P, Schonfeld A, Ovadia Y, Reisner SH. Real-time echo-Doppler Duplex Scanner in the evaluation of patients with Poland sequence. Eur J Obstet Gynecol Reprod Biol. 1989 Aug;32(2):103-8. doi: 10.1016/0028-2243(89)90190-1. PMID: 2550291.

6. Hashim, E. a. A., Quek, B. H., & Chandran, S. (2021). A narrative review of Poland's syndrome: theories of its genesis, evolution and its diagnosis and treatment. Translational Pediatrics, 10(4), 1008–1019. https://doi.org/10.21037/tp-20-320

7. Puvabanditsin S, Garrow E, Augustin G, et al. Poland-Möebius syndrome and cocaine abuse: A relook at vascular etiology. Pediatr Neurol 2005;32:285-7. 10.1016/j.pediatrneurol.2004.11.011

8. Martínez-Frías ML, Czeizel AE, Rodriguez-Pinilla E, et al. Smoking During Pregnancy and Poland sequence: Results of a Population-based Registry and a Case-control registry. Teratology 1999;59:35-8. 10.1002/(SICI)1096-9926(199901)59:1<35::AID-TERA8>3.0.CO;2-E

9. Atasoy, H.I., Yavuz, T., Altunrende, S. et al. A unique case of right-sided Poland syndrome with true dextrocardia and total situs inversus. Eur J Pediatr 172, 269–272 (2013). https://doi.org/10.1007/s00431-012-1794-5

10. Vaccari CM, Romanini MV, Musante I, et al. De novo deletion of chromosome 11q12.3 in monozygotic twins affected by Poland Syndrome. BMC Med Genet 2014;15,63. 10.1186/1471-2350-15-63

11. Baldelli, I., Gallo, F., Crimi, M. et al. Experiences of patients with Poland syndrome of diagnosis and care in Italy: a pilot survey. Orphanet J Rare Dis 14, 269 (2019). https://doi.org/10.1186/s13023-019-1253-8

12. RARE EXPERIENCE 2020 The lived experiences of people affected by genetic, rare and undiagnosed conditions. Genetic Alliance UK. 2020

13. Poland Syndrome Support and Network. https://pip-uk.org. Accessed 30/09/2024

14. Cabrera-Gonzalez M, Gutierrez-de la Iglesia D, Fontecha CG, G. Garcia R, Ortega-Centol A, Perez-Lopez LM. Poland syndrome: a new classification system based on a retrospective analysis of 74 cases. Journal of Hand Surgery (European Volume). 2022;47(10):1027-1031. doi:10.1177/17531934221101770

15. Sugarman GI, Stark HH. Möbius syndrome with Poland's anomaly. J Med Genet. 1973 Jun;10(2):192-6. doi: 10.1136/jmg.10.2.192. PMID: 4351696; PMCID: PMC1013017.

16. Puvabanditsin S, Garrow E, Augustin G, Titapiwatanakul R, Kuniyoshi KM. Poland-Möbius syndrome and cocaine abuse: a relook at vascular etiology. Pediatr Neurol. 2005 Apr;32(4):285-7. doi: 10.1016/j.pediatrneurol.2004.11.011. PMID: 15797189.

17. Muir, E. (2016). Rare Disease UK: The Rare Reality – an insight into the patient and family experience of rare disease.

18. Zurynski, Y., Deverell, M., Dalkeith, T. et al. Australian children living with rare diseases: experiences of diagnosis and perceived consequences of diagnostic delays. Orphanet J Rare Dis 12, 68 (2017). https://doi.org/10.1186/s13023-017-0622-4

19. Benito-Lozano J, Arias-Merino G, Gómez-Martínez M, Arconada-López B, Ruiz-García B, Posada de la Paz M,

et al. (2023) Psychosocial impact at the time of a rare disease diagnosis. PLoS ONE 18(7): e0288875. https://doi.org/10.1371/journal.pone.0288875

20. Anderson M, Elliott EJ, Zurynski YA. Australian families living with rare disease: experiences of diagnosis, health services use and needs for psychosocial support. Orphanet J Rare Dis. 2013 Feb 11;8:22. doi: 10.1186/1750-1172-8-22. PMID: 23398775; PMCID: PMC3599672.

21. Spencer-Tansley, R., Meade, N., Ali, F. et al. Mental health care for rare disease in the UK – recommendations from a quantitative survey and multi-stakeholder workshop. BMC Health Serv Res 22, 648 (2022). https://doi.org/10.1186/s12913-022-08060-9

22. Baldelli I, Baccarani A, Barone C, Bedeschi F, Bianca S, Calabrese O, Castori M, Catena N, Corain M, Costanzo S, Barbato GP, De Stefano S, Divizia MT, Feletti F, Formica M, Lando M, Lerone M, Lorenzetti F, Martinoli C, Mellini L, Nava MB, Porcellini G, Puliti A, Romanini MV, Rondoni F, Santi P, Sartini S, Senes F, Spada L, Tarani L, Valle M, Venturino C, Zaottini F, Torre M, Crimi M. Consensus based recommendations for diagnosis and medical management of Poland syndrome (sequence). Orphanet J Rare Dis. 2020 Aug 5;15(1):201. doi: 10.1186/s13023-020-01481-x.

23. Ruhstaller T, Roe H, Thürlimann B, Nicoll JJ. The multidisciplinary meeting: An indispensable aid to communication between different specialities. Eur J Cancer. 2006 Oct;42(15):2459-62. doi: 10.1016/j.ejca.2006.03.034. Epub 2006 Aug 28. PMID: 16934974.

24. Le Divenah A, David S, Bertrand D, Chatel T, Viallards ML. Réunion de concertation pluridisciplinaire : comment décider d'une chimiothérapie à visee palliative? [Multidisciplinary consultation meetings: decision-making in palliative chemotherapy]. Sante Publique. 2013 Mar-Apr;25(2):129-35. French. PMID: 23964537.

25. Zhang, Z. Diagnosing rare diseases and mental well-being: a family's story. Orphanet J Rare Dis 18, 45 (2023). https://doi.org/10.1186/s13023-023-02648-y

26. Baldelli, I., Baccarani, A., Barone, C. et al. Consensus based recommendations for diagnosis and medical management of Poland syndrome (sequence). Orphanet J Rare Dis 15, 201 (2020). https://doi.org/10.1186/s13023-020-01481-x

27. Associazione Italiana Sindrome di Poland. http://www.sindromedipoland.org. Accessed 26/09/2024.

28. Anderl H, Kerschbaumer S. Early correction of the thoracic deformity of Poland's syndrome in children with the latissimus dorsi muscle flap: long term follow-up of two cases. Br J Plast Surg. 1986 Apr;39(2):167-72. doi: 10.1016/0007-1226(86)90076-7. PMID: 3008899.

29. Bainbridge LC, Wright AR, Kanthan R. Computed tomography in the preoperative assessment of Poland's syndrome. Br J Plast Surg. 1991;44:604–607.

30. NiMhurchadha S, Harcourt D, Diba R, Hughes J, Smith R, Nduka C. Looking 'the same': experiences of women who have had corrective surgery for breast asymmetry. J Health Psychol. 2013 Apr;18(4):488-96. doi: 10.1177/1359105312436766. Epub 2012 Jun 11. PMID: 22689588.

31. Baldelli I, Santi P, Dova L, Cardoni G, Ciliberti R, Franchelli S, Merlo DF, Romanini MV. Body Image Disorders and Surgical Timing in Patients Affected by Poland Syndrome: Data Analysis of 58 Case Studies. Plast Reconstr Surg. 2016 Apr;137(4):1273-1282. doi: 10.1097/PRS.0000000000002018. PMID: 27018681.

32. Noisser V, Eigenberger A, Weiherer M, Seitz S, Prantl L, Brébant V. Surgery of congenital breast asymmetry-which objective parameter influences the subjective satisfaction with long-term results. Arch Gynecol Obstet. 2022 Jan;305(1):95-102. doi: 10.1007/s00404-021-06218-0. Epub 2021 Sep 3. PMID: 34480228; PMCID: PMC8782804.

33. Fodor, P.B.; Khoury, F. Latissimus Dorsi Muscle Flap in Reconstruction of Congenitally Absent Breast and Pectoralis Muscle. Ann. Plast. Surg. 1980, 4, 422–425

34. Ohmori, K.; Takada, H. Correction of Poland's pectoralis major muscle anomaly with latissimus dorsi musculocutaneous flaps. Plast. Reconstr. Surg. 1980, 65, 400–404

35. Your Guide to Breast Augmentation. Association of Breast Surgery, British Association of Aesthetic Plastic Surgeons, British Association of Plastic, Reconstructive and Aesthetic Surgeons. 2021.

36. Ellart J, et al. Asymétrie mammaire de l'adolescente et de la jeune adulte. Stabilité du résultat dans le temps. À propos de 144 patientes. Ann Chir Plast Esthet (2016),

http://dx.doi.org/10.1016/j.anplas.2016.06.003

37. Mahrhofer M, Schoeller T, Casari M, Bachleitner K, Weitgasser L. Development of A Surgical Treatment Algorithm for Breast Reconstruction in Poland Syndrome Patients Considering Severity, Sex, and BMI. J Clin Med. 2021 Sep 29;10(19):4515. doi: 10.3390/jcm10194515. PMID: 34640539; PMCID: PMC8509377.

38. Gutierrez-Ontalvilla P, Naidu NS, Blanco EL, Brito EC, Ruiz-Valls A. Autologous Fat Grafting with Percutaneous Fasciotomy and Reduction of the Nipple-Areolar Complex for the Correction of Tuberous Breast Deformity in Teenagers. Aesthetic Plast Surg. 2020 Apr;44(2):264-269. doi: 10.1007/s00266-019-01531-1. Epub 2019 Oct 31. PMID: 31673737.

39. Torre, M., Baban, A., Buluggiu, A., Costanzo, S., Bricco, L., Lerone, M., Bianca, S., Gatti, G. L., Sénès, F. M., Valle, M., & Calevo, M. G. (2010). Dextrocardia in patients with Poland syndrome: Phenotypic characterization provides insight into the pathogenesis. The Journal of Thoracic and Cardiovascular Surgery, 139(5), 1177-1182. https://doi.org/10.1016/j.jtcvs.2009.08.024

40. Fraser FC, Teebi AS, Walsh S, Pinsky L. Poland sequence with dextrocardia: which comes first? Am J Med Genet. 1997 Dec 12;73(2):194-6. doi: 10.1002/(sici)1096-8628(19971212)73:2<194::aid-ajmg16>3.0.co;2-l. PMID: 9409872.

41. Yiyit N, Işıtmangil T, Öksüz S. Clinical analysis of 113 patients with Poland syndrome. Ann Thorac Surg. 2015 Mar;99(3):999-1004. doi: 10.1016/j.athoracsur.2014.10.036. Epub 2015 Jan 27. PMID: 25633462.

42. Assadi FK, Salem M. Poland syndrome associated with renal agenesis. Pediatr Nephrol. 2002 Apr;17(4):269-71. doi: 10.1007/s00467-001-0804-z. PMID: 11956880.

43. Urschel HC Jr. Poland syndrome. Semin Thorac Cardiovasc Surg. 2009 Spring;21(1):89-94. doi: 10.1053/j.semtcvs.2009.03.004. PMID: 19632568.

44. Miller RA, Miller DR (1975) Congenital absence of the pectoralis major muscle with acute lymphoblastic leukemia and genitourinary anomalies. J Pediatr 87:146–147