Poland Syndrome: A Case of a Left Handed Infant

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Abstract
A nine month old presents with left handed preference strong enough that his mother notices. Knowing that nine months is early for hand preference, further evaluation demonstrates asymmetry of his chest wall. The child is soon diagnosed with Poland Syndrome, a rare disorder, which is usually secondary to a vascular insult in utero. It presents with a wide spectrum of chest wall, breast and upper extremity clinical manifestations. Being familiar with the normal developmental trajectories of handedness in infancy should alert a physician that preference this early is due to a weakness on the contralateral side. In this case, that knowledge allowed for diagnosis of Poland Syndrome earlier than is reported in the literature.

Introduction
In 1841, while dissecting the body of George Elt, a deceased 27 year old convict, medical student Alfred Poland described the unilateral absence of the sternocostal portion of the pectoralis major along with absence of the pectoralis minor and a hypoplastic serratus anterior and external oblique muscle. Eighty-one years later, Patrick Clarkson credited Poland with the discovery of this anatomic anomaly after examining a series of three similar cases. Poland Syndrome (PS) is thought to occur sporadically, with an unclear genetic component, due to a vascular insult occurring in the 6th week of fetal development. Clinical manifestations range from unilateral absence of the pectoralis major muscle to more extensive involvement of breast tissue as well as underlying musculature, ribs and cartilage. In this case, physical evidence of the disease was so minimal that the diagnosis was missed until the infant presented with favored use of his left upper extremity. Though many cases of PS have been described in the literature, few have presented with changes in functional status, in the setting of minimal obvious physical deformity. This case is the first described incident of PS to present with early left-handedness.

Case Report
At his 9 month old well child visit, CM’s mother made a proclamation that for the last two weeks, he seemed to prefer his left hand more than his right. Delivered by an uncomplicated cesarean section at term, his antenatal course had been normal. Until nine months of age, CM had only been seen for routine preventative visits. While physical examination revealed an asymmetry of the chest wall, both upper extremities were morphologically normal, with full range of motion.

Understanding that children do not develop handedness until two years of age, he was referred to Neurology, who felt that he might have Cerebral Palsy, despite the lack of spasticity. A chest x-ray and MRI of the brain were obtained. Finding no neurologic abnormality, he was referred to Orthopedic Surgery, where he was given the diagnosis of Poland syndrome after detecting an absent sternocostal portion of the pectoralis major and pectoralis minor. The defect was thought to be cosmetic and therefore not needing repair. Further imaging indicated no structural malformation to his right sided ribs. Position of internal organs were also normal. His left handedness remained for the next two years before gradually shifting to equal hand utilization. Before his fourth birthday, he solidified right hand preference. Currently, at nine years old, despite still displaying a subtle asymmetry in is chest wall (Figure 1), CM is a very athletic child, who has shown no physical limitations.

Discussion
Poland Syndrome (PS) is rare, occurring in 1 in 30,000 live births, but ranges of 1 in 7,000 to 1 in 100,000 are quoted within the literature. PS is defined as a unilateral malformation of the sternocostal head of the pectoralis major with ipsilateral malformations of the anterior chest wall and breast. Predominately in males (3:1), it occurs on the right side between 60-75% of the time. It is usually sporadic, but a rare Autosomal Dominant familial inheritance pattern with
incomplete penetrance exists. A referenced occurrence in only one sibling of a set of monozygotic twins dispels pure genetic transmission.  

The phenotypic pathology of PS is variable but nearly always ipsilateral. Chest wall anomalies must include the pectoralis major abnormalities listed above, but can include the pectoralis minor, serratus anterior, external obliques, infraspinatus, latissimus dorsi and deltid. Aplasia or deformities of the ribs and costal cartilages, particularly 2nd to 4th or 3rd to 5th ribs, can be seen. Hypoplasia of the breast or areola can be present and subcutaneous fat can be deficient. Alopecia and anhidrosis of the axillary and mammary regions can be involved. Upper extremity malformations, if they exist, can range from webbing of the fingers (syndactyly) to shortened upper arm, forearm and fingers (brachysymphalangism) to a phocomelia-like deficiency.  

Rarer reported associations with PS are Mobius syndrome (congenital bilateral facial nerve palsy), Klippel-Feil syndrome (congenital fusion of any two cervical vertebrae and brainstem or cerebellar abnormalities), X-linked Ichthyosis (steroid sulfatase deficiency), Parry Rhomberg syndrome (acquired progressive hemi-facial palsy) and Sprengel deformity (absence of upper portion of the Serratus anterior or ‘winged scapula’). Other affected systems that have been reported including cardiac (dextrocardia), GI (situs inversus, hiatal hernia), neurologic, dermatologic (Cafe-au-lait spots, neurofibromas, melanocytic nevi,
hemangiomas and psoriasis) and renal (hypoplastic kidney).\textsuperscript{2,4,5} Cancers associated with PS have included Leukemia, non-Hodgkin lymphoma, Leiomysarcoma, lung cancer, phyllodes tumor, invasive ductal carcinoma of the breast and Wilms’ tumor.\textsuperscript{5}

The primary theory behind PS development is Subclavian Artery Supply Disruption Sequence (SASDS), in which a vascular disruption occurs in the sixth week of development causing hypoplasia at some point along the Subclavian artery or its branches.\textsuperscript{1,2,3,4,5} The thoracic wall is supplied by branches of the axillary artery (medial thoracic, intercostals and thoracic arteries), the thoracoacromial artery and the lateral thoracic artery. All arise from the subcapsular and axillary arteries, which originate from the subclavian artery (Figure 2).\textsuperscript{4} Findings of vascular hypoplasia indicate vascular etiology.\textsuperscript{2} The sixth week of gestation is when the pectoral mass splits into costal and clavicular heads. Furthermore, digits begin to form as digital rays.\textsuperscript{2,5} How much and in which locations the artery and branches are disrupted account for the widely varied presentation.\textsuperscript{4}

Other theories for development of PS include disruption of the lateral embryonal plate mesoderm after 16-28 days post fertilization, atrophy of the cord anterior motor neurons, intrauterine trauma, viral infections and teratogens.\textsuperscript{2,5}

In this case, the patient was noted to have a hand preference strong enough for a parent to identify a preference by the ninth month. The development of a functioning reach in an infant requires Straightness, which is kinematically defined as the ratio of the path traveled by the hand over a distance to the target, and Smoothness, which is a lack of vacillations between accelerations and decelerations along that path.\textsuperscript{7} Unless there is a unilateral cerebral palsy,\textsuperscript{8} even 2D and 3D motion analysis cannot determine a difference in either Straightness or Smoothness between hands until at least 9 months.\textsuperscript{7} In fact analysis of pre-reaching movements (head turning or post-startle preferences) in the first months of life show no correlation with future handedness.\textsuperscript{9} The difficulty in assessment of the development of handedness is that periodic standard assessment of developmental milestones are not able to detect change and standard neurologic exams have a poor coorelation.\textsuperscript{8}

Overall, there is a human right-handed bias. That can be seen when the development of handedness is analyzed longitudinally. The infants who eventually develop right-handedness, have a more homogenous development trajectory than do eventual non-right hand dominant (left handed or ambidextrous) infants. Even these non-right hand dominant infants would occasionally demonstrate periods or right-handedness.\textsuperscript{10} With regular intensive evaluations, trajectories begin to lateralize from twelve to eighteen months.\textsuperscript{9}

Still, there is fluctuation of these trajectories based on a complex cascade of developmental contingencies. Bimanual manipulation requires acquiring objects with the non-dominant hand, then allowing the dominate hand to manipulate it. The change in positional orientation of the developing child’s head from supine to upright, or congenital postural asymmetry, impacts hand usage. Even interaction with parents, most of whom will be right handed, will impact development.\textsuperscript{10} At two years of age however, most children have a constant manual preference.\textsuperscript{9}

Clearly, until handedness is solidified, it is a vacillating process evidenced by Michel and Babik’s work, where some infants had different trajectories whether odd months or even months were extracted for analysis.\textsuperscript{10} For a mother to notice her infant’s hand preference at nine months, with the highly fluctuating nature of development of handedness, there needs to be a cerebral deficit, or in this case, a musculoskeletal weakness on the non-dominant side.
Conclusion

This case illustrates how PS can present in the mildest of cases and how some functional impairment of the ipsilateral limb can be seen clinically, even in cases without associated hypoplasia of the upper extremity. Knowledge of developmental trajectories of handedness in the first two years of life can allow for earlier identification.

Acknowledgements

We would like to thank Dr. Tammy Bannister and Joshua Botkin for their contributions, which were essential in this case study.

References

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