

SWISS SOCIETY OF NEONATOLOGY

Moebius syndrome with
Poland anomaly

DECEMBER 2005



Stritzke A, Pasquier S, Zeilinger G Neonatal Intensive
Care Unit, Kinderklinik Aarau, Switzerland

Congenital facial diplegia or plegia, Diplégie faciale congénitale, Diplegia facial congénita, Möbius syndrome, Nuclear hypoplasia congenital, facial diplegia, Akinesia algera, congenital abducens-facial paralysis, congenital bulbar paralysis, congenital facial paralysis, congenital nuclear agenesis, congenital nuclear aplasia, congenital oculofacial paralysis, congenital paralysis of the sixth and seventh nerves, infantile nuclear aplasia, nuclear agenesis syndrome, oculofacial paralysis syndrome; Syndactylie de type Poland, Sindactilia de Poland, Symbrachydactyly with ipsilateral aplasia of sternal head of pectoralis major muscle, Poland syndactyly

SYNONYMS

This female infant was born at 40 6/7 weeks of gestation after an uneventful pregnancy to a 30-year-old mother and a 38-year-old father without consanguinity who, as well as their two older children, born in 1999 and 2001, were in good health condition. Delivery and adaptation were normal, growth parameters were within normal limits, birth weight being 3450 g, length 53 cm and head circumference 35 cm.

CASE REPORT

The following clinical findings were noted: Amastia and undefined anterior axillary fold on the left side indicating malformation of the sternal head of the major pectoral muscle, connatal paralysis of the left facial nerve with normal pupillary reaction and residual function to close the eye. Slight deviation of the

tongue to the left side indicating a partial paralysis of the hypoglossal nerve (Fig. 1-4). On the left hand partial cutaneous syndactyly of the index and middle finger and klinodactyly Dig V, brachydaktyly Dig II with nail hypoplasia Fig. 5, 6). Strabismus convergens and an abnormal space between Dig I and II of both feet were noted. On x-ray brachydactyly and shortening especially of the middle phalanges, but also partly the distal phalanges and thumb were diagnosed. A small atrial septal defect was demonstrated on echocardiography.

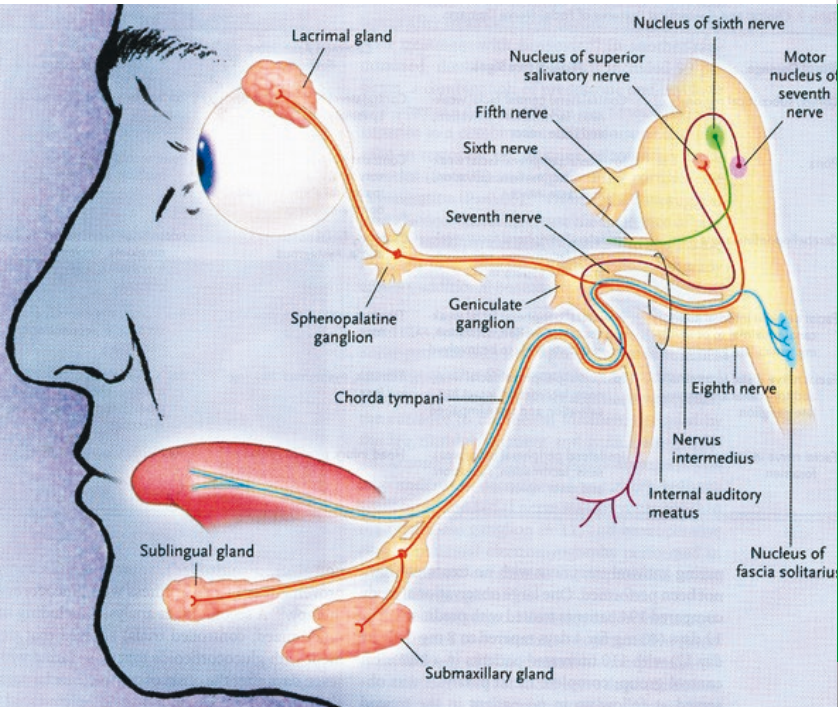


Fig. 1

Anatomy of the facial nerve (courtesy of BMJ 2004).



Fig. 2

Amastia and aplasia of sternal head of major pectoralis muscle on the left side.



Fig. 3

Left facial paralysis in a newborn girl with Moebius and Poland syndrome.



Fig. 4

Left facial paralysis in a newborn girl with Moebius and Poland syndrome.

**Fig. 5**

Simple cutaneous syndactyly Dig III/III, clinodaktyly Dig V on the left hand.



Fig. 6

For comparison, normal right hand.

Moebius syndrome has been recognized as an entity since 1888 (Original: P. J. Möbius: Über angeborene doppelseitige Abducens-Facialis-Lähmung. Münchener medizinische Wochenschrift 1888;6:108-111). According to one author it was first described by von Graefe in 1880. However, since Albrecht von Graefe died in 1870, this probably refers to the textbook by Alfred Graefe and Edwin Theodor Saemisch. It was described by Möbius in 1888 and again in 1892 when he reported 44 cases and applied the term «nuclear atrophy». Moebius syndrome is defined as nonprogressive, congenital syndrome characterized by paralysis of the 6th and 7th cranial nerves. Facial features include usually bilateral facial paralysis, with incomplete paralysis affecting the upper face more than the lower, weakness of facial muscles including orbicularis oculi muscles with mask-like face and inability to smile due to lack of facial expression, ocular ptosis and open eyes during sleep, inability to close the mouth while chewing as well as convergent strabism due to abducens palsy.

The abducens palsy also tends to be bilateral and complete, affecting approximately 75% of the patients having paralysis of the lateral rectus muscle. External ophtalmoplegia has been reported in 25%. Atrophy of tongue due to lack of hypoglossal innervation, drooling, weakness of palate and external ear deformities with occasional hearing loss may be noted. Other clinical features may include occasio-

nal mental retardation of a moderate degree (10%), skeletal or muscle deformities as talipes equinovarus, congenital amputations and Poland anomaly. Poland syndrome is named after Alfred Poland, demonstrator in anatomy and later surgeon and ophthalmologist at Guy's Hospital in London where he dissected the body of a 27-year-old deceased convict named George Elt in 1841 whom he reported had „Deficiency of the pectoral muscles“ (Guy's Hosp Rep 1841;6:191).

Poland anomaly has two major components: unilateral aplasia of the sternal head of major pectoralis muscle and ipsilateral symbrachydactyly. Clinically there is absence of the normal anterior axillary fold, the clavicular head of the pectoral muscle on the other hand is always present and sometimes hypertrophied. Symbrachydactyly consists of shortening or even absence of digits associated with tissue webbing of variable degree, frequently involving the index and middle fingers. Middle phalanges are affected more frequently, they may be absent or fused with the distal phalanges (terminal symphalangis and assimilation hypoplasia). Distal phalanges are minimally affected and rarely absent. The thumb is usually least affected. Asymmetry of breast development with ipsilateral absence of breast and subcutaneous tissue, as well as webbing of the axilla may be additional findings.

Cases seem to manifest sporadically in about 1 in 20'000 children, rarely with dominant autosomal

inheritance, rather probable is etiological heterogeneity. Etiology and pathogenesis is unknown, in some cases intrauterine vascular compromise, i.e. embryological disruption of subclavian artery development has been suspected. Agenesis or early disruption of development of motoric ganglion neurons in the brain stem are postulated. Hypoglossy-Hypodactyly as well is another common feature. No effect on intelligence and normal development has been noted. Poland syndrome has been found to be more common in males (3:1), the right side is more commonly involved than the left (2:1). Recurrence risk is low.

Other anomalies in association with Poland syndrome are: Radioulnar synostosis, Sprengel deformity, coalition of the carpal bones, camptodactyly, polydactyly, skin dimples, deficiencies of the rib cage, scoliosis, kyphosis, cervical ribs, club foot, metatarsus adductus and syndactyly of the toes. Visceral anomalies include: dextrocardia, herniation of the lungs, inguinal and umbilical hernias, cryptorchidism, ipsilateral hypoplasia of the kidney, encephalocele and microcephaly. Diagnosis is usually made upon characteristic dysmorphias at birth, there is no diagnostic laboratory test. Electromyographic studies are usually abnormal with few or absent motor unit potentials tending to verify a supranuclear or nuclear cause for the palsies. The basic defect accounting for cranial nerve palsy is generally unknown, nuclear hypoplasia is documented in a few cases of the approximately 140 reported

cases with occasional familial aggregation. Autosomal dominant transmission occurs. Frequency of consanguineous marriages is a feature of many reported patients.

Prognosis is usually compatible with a normal life span and there is no progression in muscle weakness from infancy to adulthood. Problems may include feeding, drooling of saliva, swallowing, aspiration and indistinct speech at a later age. Failure to thrive, corneal and conjunctival ulceration and aspiration bronchopneumonia are complications. Care is general symptomatic with protection of the eyes against exposure keratitis and surgical correction in case of syndactyly. There also has been proposed operational approaches with institution of muscle fibers as the gracilis muscle with direct repair of the muscle's motor nerve to the masseteric branch of the trigeminal or other facial nerve to reconstitute patients mimical ability. Long-term outcome and wide-spread use of this technique is lacking.

1. Atlas of the face in genetic disorders, Richard M. Goodman, Robert J. Gorlin, Second Edition, 1977
2. Principles and practice of medical genetics, Volume 2, Alan E.H. Emery, David L. Rimoin Editors, Churchill Livingstone 1983
3. Birth defects compendium, Second Edition, Daniel Bergsma, Published for the National Foundation, March of Dimes by the Macmillan Press Ltd, 1979

REFERENCES

SUPPORTED BY



CONTACT

Swiss Society of Neonatology
www.neonet.ch
webmaster@neonet.ch