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Eugène Apert and his Contributions to Plastic Surgery

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Abstract

French pediatrician Eugène Apert is best known for his 1906 description of the eponymous Apert's Syndrome: the widely recognized congenital condition that is known as acrocephalosyndactyly, which is characterized by distinct craniofacial deformities and bilateral syndactyly of the hands and feet. Subsequent efforts to study and treat this condition have led to contributions from numerous medical and surgical specialties under the guidance of plastic surgery. Apert's influence on medicine, however, extends far beyond what can be appreciated by the impact of his eponymous syndrome. Considered one of France's eminent pediatricians, Apert additionally made important contributions to the study of adult diseases. He was also a founding member of the French Eugenics Society, serving as its secretary general and president in a tenure that lasted for most of his career. Apert's medical contributions within the context of this scientific ideology make him an important and potentially controversial figure in medicine.

Keywords

Eugène Apert; Apert's syndrome; acrocephalosyndactyly; French eugenics; historical perspective

A renowned figure in medicine during his time, Eugène Apert (1868–1940) is perhaps best known today for describing the syndrome acrocephalosyndactyly that is synonymous with his name. With its classic craniofacial deformities and bilateral complicated syndactylies of the hands and feet, Apert's syndrome is a condition instantly recognizable by clinicians. Nevertheless, history has a way of forgetting the person behind the eponym and disregarding the lesser well-known contributions. Apert is no exception. A closer look at his work reveals the scope of his impact being broader than what is generally appreciated today, and raises questions as to the influence of his involvement in the controversial branch of science, eugenics.

Although Apert wrote prodigiously throughout his career, a PubMed search failed to return any articles written by the French physician. At the time of this writing, only one article exists in the PubMed database explored Apert's contributions to medicine [1]. Search efforts thus relied on archives from the University of Michigan Taubman Medical Library and

loaned collections from other libraries. Because these original books and manuscripts presented by Apert were published in French, an exhaustive translation of his seminal work was conducted to understand the impact of his contributions. Key findings included several of his original articles, including his 1906 description of acrocephalosyndactyly that would come to bear his name, countless medical texts he authored in the early 1900's, and numerous obituaries written by his former students.

Early Career and Initial Description of Apert's Syndrome

Apert received his M.D. from *Faculté de Médecine* in 1897, completing his thesis on purpuras in the same year [2]. He then went on to work under the mentorship of several distinguished physicians, among whom included the surgeon Paul Georges Dieulafoy, pediatrician Jacques-Joseph Grancher, and obstetrician Pierre-Constant Budin [3]. His time with the latter two in particular laid the ground for his interest in pediatrics. He further cemented his expertise in childhood illness as a student and occasional collaborator of the renowned Antoine Bernard-Jean Marfan. Following his appointment as *medecin des hopitaux* (Doctor of Hospitals) in 1902, Apert continued to devote himself to pediatrics, working in various children's hospitals throughout France [4].

In 1906, Apert published "De l'acrocephalosyndactylie", his now famous description of patients with a unique set of craniofacial, hand, and foot deformities. He proposed a single clinical entity:

I suggest the name, acrocephalosyndactyly to designate a type of teratology compatible with life and strongly characterized by the coexistence of the two following particularities: First, a high skull flattened in the back and sometimes at the sides, bulging on the opposite side to an exaggerated facet in the superior frontal region; second, syndactyly of the four extremities [5].

Interestingly, Apert's study included only one patient that he had examined himself in 1898 when still an intern. Much of this one case report contained a review of 8 other cases he had amassed from the literature. Apert diligently cited others, giving credit for the earliest description to fellow French physician Trouqart, who in 1886 described a newborn with identical features [6]. Apert consolidated cases like this and notes from his own observations to provide an excellent overview of the morphological characteristics.

Proceeding in a systematic fashion, Apert meticulously described every deformity. Take for example his descriptions of the craniofacial and hand anomalies:

The skull presents in the following fashion; it is flattened on the posterior face of such sort that the bulge of the occiput is no longer there and is replaced by a flattened surface that is found on the same level plane as the posterior surface of the neck. The occipital bone is flat and vertical; it isn't but beyond the posterior fontanel that the contour of the skull inclines in the front continuing upward. The most elevated part of the skull corresponding to the anterior fontanel is found nearly on the same plane as on the forehead and the face...

The hands are also affected by syndactyly, carrying at the least four internal digits united in a single mass; the union is the most intimate at the extremity of the fingers; most of the nails of the three median fingers are joined into a single one; sometimes even the nail of the little finger is surrounded in an enormous quadruple nail. The palm of the hand is immobilized in half flexion and the form of the hand is compared to that of a spoon, or of that of a goose's head. The thumb is not ordinarily also completely fused, but it is often immobilized more or less and joined, directed transversely [5].

Apert described a similar bilateral syndactyly observed in the feet of these patients. In addition, he noted a spectrum of additional craniofacial abnormalities, including widening of the anterior sutures, bulging of the eyebrows, atrophied maxillae, depression of the nasal bones, and palatal malformations.

Using a combination of anatomical, artistic, and practical terminology, Apert managed to convey vivid depictions of these complex deformities. This may simply be a necessity because of a lack of pictures or artists' renditions, but the clarity and simplicity in which he presented each case reveals his astute observations. Complementing exacting measurements with simple directional and geometric references, Apert struck an elegant balance between providing a high degree of detail and minimizing the use of complex language. Apert also discussed what several investigators had postulated as the etiology of this syndrome, namely fetal syphilis, amniotic compression, and abnormal extremity development secondary to skull deformities interfering with pituitary function. However, the discussion on these matters was brief because Apert's primary goal was "only to establish the particular morphological type of acrocephalosyndactyly." His detailed descriptions have withstood the test of time, and remain the foundation for current clinical diagnostic standards. Slight modifications to these criteria are the result of advanced imaging, surgical, and autopsy studies, and only refine what Apert initially described. This includes the observed complete synostosis of the coronal sutures as well as the extent of bony fusion in the extremities [7, 8].

In 1920, Park and Powers performed the first comprehensive evaluation of Apert's proposed acrocephalosyndactyly [9]. In their exhaustive review of the literature, they reported on 30 cases from 8 different countries with similar deformities. While establishing acrocephalosyndactyly as not extremely uncommon, Park and Powers were reluctant to attribute the craniofacial and extremity anomalies to a common cause, citing the extreme variation in clinical presentation, and the well documented occurrences of isolated identical deformities. Their assessment effectively rejected Apert's notion of his acrocephalosyndactyly being a single clinical entity, stating, "Accordingly we have felt obliged to discard the title, 'acrocephalosyndactylism'." However, 40 years later, Blank provides the next significant assessment in his review of 54 cases to support Apert's view that this syndrome is indeed caused by a single etiology. His work was the first to rely predominantly on first-hand observation (34 out of 54 patients were available for examination), rather than descriptions from the literature. In addition to discussions on incidence, and the possible effect of parental age, he attributed the syndrome to a single gene in heterozygous form. Blank then proceeded to divide his cases into two main clinical

categories: typical acrocephalosyndactyly (Type I) as originally described by Apert, and atypical acrocephalosyndactyly (Type II) which lacks the complete bilateral syndactyly universally seen in typical cases. More importantly, with this categorization, the term “Apert’s syndrome” was first used in reference to the typical acrocephalosyndactyly. Perhaps in response to Park and Powers’ assessment 40 years earlier, Blank further states “Apert’s syndrome and atypical acrocephalosyndactyly are probably etiologically unrelated.” [10] Support is thereby given to Apert’s initial assessment and at the same time still giving due credit to Park and Powers for their meticulous work.

Impact of Apert’s Syndrome on Medicine

The subsequent impact of Apert’s syndrome on medicine can be appreciated by the myriad of medical fields involved in its management. In true multidisciplinary fashion, specialists from all branches of medicine are consulted to treat this disease. Geneticists provide the initial diagnosis and counseling, followed by neurosurgeons who evaluate for fusion of the cranial sutures and hydrocephalus. Pediatricians assume a crucial role in maintaining general health during the formative years, as do otorhinolaryngologists, ophthalmologists, audiologists, and psychologists [7]. Plastic surgeons play an especially important role in the management of Apert’s syndrome, as its complex craniofacial, hand, and foot deformities present unique functional and cosmetic challenges. In this context, one sees the effects of Apert’s work surfacing at key milestones in the evolution of sub-specialties within plastic surgery.

In 1967 at the International Plastic Surgery Congress in Rome, French surgeon Paul Tessier established himself as the father of craniofacial surgery by presenting a series of cases that summarized his approach for treating craniofacial deformities [11, 12]. In particular, his techniques for correcting hypertelorism and mid-face retrusion – two of the more cosmetically displeasing and potentially medically concerning facial features of Apert’s syndrome – were considered landmark contributions in plastic surgery. For hypertelorism, he presented a novel intracranial approach that avoided insult to the eye and brain. For mid-face retrusion, he applied osteotomies that reproduced the classic Le Fort III fracture lines [13]. This latter work in particular relied heavily on his experience with Apert’s syndrome patients [11].

Tessier subsequently continued to revise and add to the repertoire of techniques available to treat Apert’s syndrome. The seemingly overwhelming number of surgical options is best explained by the surgeon himself, who stated “Treatment cannot be uniform since the basic anomalies vary considerably, depending on phenotype, age, and whether there have been previous craniotomies. Therefore the surgical tactics and procedures must be flexible”. As such, he followed up with recommendations in treating the frontal, orbital, maxillary, and mandibular anomalies of Apert’s syndrome. Moreover, Tessier considered Apert’s syndrome the prototype and most severe form of acrocephalosyndactyly [14]. His assessment reflects the syndrome’s technical challenges, and how lessons learned from its treatment could be applied to the management of other craniofacial deformities. Clearly, the work that established the field of craniofacial surgery relied in no small part on the syndrome Apert initially described.

Hand surgeons have also long been involved in treating the complex bilateral syndactyly universally seen in patients with Apert's syndrome. However, it was not until 1970 that a study by Hoover addressed the surgical management of the syndactyly specific to Apert's syndrome. He recommended using a modification of Bauer's zigzag technique [15] with a dorsal rectangular flap in a two-step process; the first step involving release of the thumb and little finger before age 2, followed by release of the index, long, and ring fingers 6–9 months thereafter. [16]. Hoover's work then set the stage for other astute hand surgeons to consider the complex interplay between the anatomical, developmental, and psychosocial issues unique to the Apert hand. Notably, in 1986 Barot and Caplan advocated for even earlier syndactyly release at 7–8 months [17]. More recently, different methods for flap coverage, as well as ways of improving hand function beyond what syndactyly release can offer have been proposed. Emphasis has also been placed on preservation of all 5 digits, as a lack of fingers has been noted to be psychologically distressing, particularly in younger patients [18–20].

Other Contributions

Only 38 when “De l'acrocephalosyndactylie” was published, Apert was in fact at the very beginning of a long and distinguished medical career. World War I found him fulfilling his patriotic duty as a military physician at *Andral* Hospital until 1919, when he received a senior appointment at the *Hopital des Enfants-Malades* (Sick Children's Hospital) [3]. It was here that he stayed for the remainder of his career and established himself as a pediatric expert. Unknown to many today, this was also the time of Apert's involvement in a controversial scientific ideology: eugenics.

It would not be unreasonable to assume that the seedlings of Apert's interest in eugenics came from his experience with childhood illness, particularly of the inherited type. A brief review of his publications on the subject is quick to prove the point. Starting with a book he co-wrote with Dieulafoy in 1907 entitled Traits of Familial and Congenital Diseases [21], he went on to write Morbid Heredity in 1919 [22]. This was followed by Eugenics and Selection in 1922 [23], and The Twins: A Study of their Biology, Physiology, and Medicine in 1923 [24]. Apert's interest in genetics and its role in the pathogenesis of inheritable diseases were evident from these texts, but his driving motivation was best demonstrated by a speech he gave to the all female French Red Cross personnel in 1913. This speech “The Problems of Heredity”, offered a fascinating glimpse into his scientific and social thinking. In his address, he celebrated Mendel's recently re-discovered work, and its application to the inheritable diseases seen in humans – a concept considered novel at the time. He explained with impressive simplicity Mendel's theories of inheritance, and supported these theories with practical clinical observations. He discussed similar patterns noted in diseases, acquired congenital anomalies, as well as “moral characteristics” and “intellectual faculties.” In a rather amusing attempt to prove the latter point, Apert pointed to the Bach, Darwin, and Galton family lineages with their impressive musical and scholastic accomplishments. Moreover, Apert acknowledged that inheritance alone did not account for all of the observed patterns, and that other factors played a role. He then concluded his speech with several points of practical importance, the last of which being most revealing, in which he states, “We can hope that these efforts will show us the way to secure amelioration for the

generations to come, and a lessening of congenital defects, the frequency of which weighs so heavily upon poor humanity” [25].

Apert continued to play a key role in the French eugenics movement, lending an authoritative voice and serving as a legitimate link to the medical community. He went on to help found the French Eugenics Society that same year and served as its first secretary general, and later as president [26]. Throughout a tenure with the society that lasted until his death, he was a staunch believer in using eugenics as a means to improve the national health of France, particularly in the aftermath of World War I [26]. In his paper, “Eugenics and National Health,” Apert described an increased frequency in the number of birth defects observed in children conceived during the war. He further described the increased post-war incidence of syphilis and tuberculosis amongst children and women, as well as the deleterious implications of wartime malnutrition on the young. Apert recognized these trends as having immediate and long-term consequences on French national health, and advocated the need for increasing the birthrate, improving sanitation, better working conditions, and targeted health care programs [27].

There are aspects of Apert’s eugenics work however, that when viewed through a historical lens appear as shades of racism. Following World War I, Apert and others grew concerned over the increasing number of immigrants from other countries making their way into France. They believed that an influx of immigrants would introduce deleterious genetic elements into the French population. At the 1922 French Eugenics Society meeting, Apert himself voiced concern over the increasing number of “mulattoes” being born in an area of southern France where a Vietnamese Senegalese battalion was stationed. The following year, he suggested banning children of black or Asian descent from entering France [26]. Just prior to his death in 1940, Apert again emphasized the need to regulate immigration in his overview on the state of eugenics in France, throwing his support behind regulations to ensure the genetic fitness of foreigners [26, 28]. That Apert supported such racially charged measures is surprising, particularly in regards to his stance on banning certain ethnicities. Moreover, one wonders how the French pediatrician justified his abandonment of children of supposedly inferior race. At the very least, his judgment reflects a racism that was not uncommon in the thinking of his time. It is disconcerting to appreciate Apert’s work on behalf of children, when at the same time, his eugenic views were exploited by Nazism in the guise of better good for humanity.

Despite his eugenicist beliefs, Apert was unequivocally regarded as a doctor of children. Ever the sensitive caregiver, his kind and simple bedside manner belied his clinical acumen [4]. He published on a wide variety of pediatric subjects, including mental retardation, infantilism, growth, and hygiene. [29–34]. Considered France’s leading pediatric expert at the height of his career, his “Manual on Hygiene for the First Years” was considered the definitive guide on raising children by French mothers [4]. Apert also took seriously his role in contributing to general medicine, writing on a variety of adult diseases and pathology [2, 35, 36], including the first description of hirsutism caused by corticoadrenal tumors [37].

Apert the Man

Revered by patients and students alike, Apert was described as a thoughtful, almost reserved man of character who spoke with a gentleness that enamored him to anyone he came across. Interns were always grateful to learn from the “master” as his tall figure strode from bed to bed, patiently and tenderly assessing each child. Never one to seek recognition, he rejected – albeit gratefully – the notion to have a retirement party in his honor [38]. Here then, is a man who remained the modest physician, taking only the satisfaction from caring for his young patients, teaching his eager students, and ensuring the national health of his beloved France. In this last context, his involvement in eugenics also reveals a man of contrast; a man who for all his altruistic work as a pediatrician, conformed to the racist sentiments of the time. Regardless, Apert’s influence on medicine is undeniable. From his eponymous syndrome to his contributions to pediatrics, his work has transcended both time and medical specialties, and is a testament to his impact as a physician.

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Figure 1.
Eugène Apert. Taken from: Cambessédès, H., *Eugène Apert (1868–1940)*. Paris Medical, 1940. **30**: p. 235–236.