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A Bibliometric Analysis of Clinical Geneticists Pioneering Clinical Genetics in the Developing World

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Abstract

Background: Clinical genetics has advanced significantly due to the contributions of pioneering physicians, particularly in developed countries. However, less is known about the key figures shaping the field in the developing world.

Materials and Methods: This study aims to identify and evaluate the impact of clinical geneticists from 174 developing countries using bibliometric analysis. Over 1,000 Google Scholar Citation profiles were analyzed in January 2025, with a focus on individuals who have an H-index of 20 or higher, indicating significant scholarly impact.

Results: The results revealed notable clinical geneticists from countries including Iraq, China, Iran, and India. The study also identified regions with limited academic representation, highlighting global disparities in clinical genetics research and practice. Notably, Aamir Jalal Al-Mosawi from Iraq emerged as a key figure in the field, making groundbreaking contributions to genetic medicine in the region.

Conclusion: The findings underscore the importance of enhancing investment in clinical genetics research and education, particularly in underrepresented regions, to address global health disparities and foster broader advancements in genetic medicine.

Keywords: Clinical Geneticists; Pioneers

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Introduction

Clinical dysmorphology, the study of congenital malformations and syndromes, is considered an integral component of clinical genetics. Early milestones in clinical genetics trace back to foundational discoveries in inheritance, beginning with Gregor Mendel's work on genetic inheritance patterns in the mid-19th century (Figure 1A). Mendel's pioneering experiments with pea plants set the stage for future advancements in genetics.

William Bateson (Figure 1B), a key figure in the development of genetics, was the first to coin the term "genetics" in 1902. Bateson, a strong advocate of Mendel's principles, published a book titled Mendel's Principles of Heredity: A Defense (Figure 1C), further establishing the scientific foundation for the emerging field of genetics.

Clinical genetics continued to evolve through the contributions of numerous pioneers who helped identify and describe genetic disorders.

One of the earliest recognized genetic conditions, Down syndrome (Trisomy 21), was initially described by Jean-Étienne-Dominique Esquirol (Figure 1D) in 1838, and later by Édouard Séguin (Figure 1E) in 1846.

However, the disorder was named after John Langdon Down (Figure 1F), a British physician who emphasized that the syndrome is a distinct form of mental retardation in 1862. The condition was later identified as a chromosomal disorder, specifically trisomy of chromosome 21, by Dr. Jérôme Lejeune (Figure 1G) in 1959, which led to the widely accepted term "Trisomy 21" [1].

Another early example of genetic disorders described in the 19th century is muscular dystrophy. Giovanni Semmola (1793-1866), an Italian physician, is considered one of the first to describe a severe form of early-onset muscular dystrophy in 1834. Semmola documented two cases of boys with muscular dystrophy exhibiting marked muscular hypertrophy and presented his findings in a lecture to Academia Pontaniana in Naples.

Figure 1A: Gregor Johann Mendel (20 July 20, 1822-January 6, 1884), an Austrian biologist.

Figure 1B: William Bateson (August 8, 1861-February 8, 1926), an English biologist.

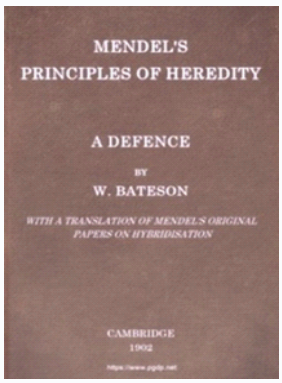

Figure 1C: William Bateson' book entitled " Mendel's Principles of Heredity: A Defense."

Figure 1D: Jean-Étienne Dominique Esquirol (February 3, 1772-December 12, 1840), a French psychiatrist.

Figure 1E: Edouard Séguin (January 20, 1812-October 28, 1880), a physician who was best known for his work with children with cognitive impairments in France and the United States.

Figure 1F: John Langdon Down (November 18, 1828-October 7, 1896), a British physician.

In 1852, Dr. Edward Meryon (Figure 1H) reported a familial condition affecting males that caused significant muscle degeneration without affecting the central nervous system. Meryon noted that the disorder led to early muscle degeneration, often with fatty infiltration. He suggested a genetic basis for the disease, transmitted through females, which affected males specifically.

In a report to the Royal Medical and Chirurgical Society in December 1851, Meryon detailed his observations of eight boys from three families, contributing significantly to the understanding of this condition. His communication was published in the Transactions of the Society in 1852. Additionally, in his 1864 book Practical and Pathological Researches on the Various Forms of Paralysis (Figure 1I), Meryon elaborated on his findings and included two additional families with similar presentations.

In 1861, the French physician Guillaume-Benjamin-Amand Duchenne provided a comprehensive description of what is now known as Duchenne muscular dystrophy. Duchenne named the condition progressive muscular atrophy and, in 1868, conducted the first muscle biopsy on an affected patient, advancing both diagnostic and treatment approaches for muscular dystrophy. In his 1861 work Paraplegie hypertrophique de l'enfance de cause cérébrale, Duchenne included a detailed case study of a young boy with the condition. His 1862 Album de photographies pathologiques, an atlas of pathological photographs, featured images of his patients. Duchenne's examination of tissue biopsies was a pioneering technique in clinical genetics [2].

In the early 20th century, Cornelia C. de Lange (Figure 1J), a Dutch

Figure 1G: Jérôme Jean Louis Marie Lejeune (June 13, 1926-April 3, 1994), a French pediatrician and geneticist.

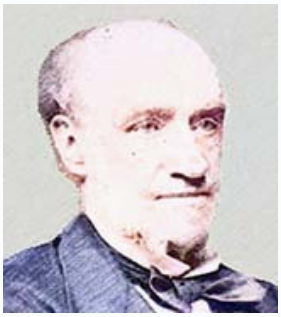

Figure 1H: Edward Meryon, an English physician.

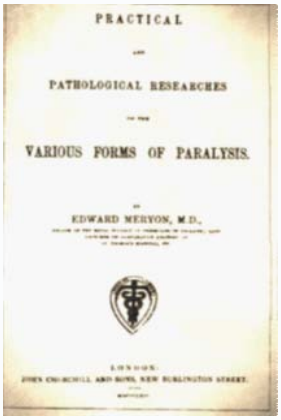

Figure 1I: The book entitled practical and pathological researches on the various forms of paralysis published by Edward Meryon.

pediatrician, made significant contributions to clinical genetics, particularly in her identification of Cornelia de Lange syndrome, a rare genetic disorder. Her work continues to have a lasting impact on the field of dysmorphology and genetic syndromes [3].

The 20th century also witnessed the development of a more systematic approach to understanding inherited conditions.

Victor A. McKusick (Figure 1K), considered the father of modern clinical genetics, is renowned for establishing the Mendelian Inheritance in Man (MIM) database, later expanded into the online resource Online Mendelian Inheritance in Man (OMIM). McKusick's work laid the foundation for much of contemporary clinical genetics, earning him recognition as a key figure in the field's development [4, 5].

Figure 1J: Cornelia C. de Lange (1871-1950), a Dutch pediatrician pioneered the field of clinical genetics during the first half of the twentieth century.

Figure 1K: Victor Almon McKusick (October 21, 1921- July 22, 2008), an American physician.

Materials and Methods

This study employed a bibliometric analysis to identify clinical physician leaders who have pioneered clinical genetics in the developing world. Biologists and laboratory geneticists were excluded from the study, as the focus was specifically on clinical geneticists. Over 1,000 Google Scholar Citation profiles were examined in January 2025, with a focus on individuals from 174 developing countries. The inclusion criterion was an H-index of 20 or higher, which reflects significant scholarly impact in the field.

Results

The analysis identified notable clinical geneticists from several developing countries, including:

- Aamir Jalal Al-Mosawi [Iraq], H-index 24 [6]
- Nikoletta Nagy [Hungary], H-index 24 [7]
- Yang Xinping [China], H-index 23 [8]
- George A. Tanteles [Cyprus], H-index 22 [9]
- Mehdi Dianatpour [Iran], H-index 21 [10]
- Erwin Brosens [Bulgaria], H-index 20 [11]

Additionally, several countries had clinical geneticists with H-indices of 10 or higher, indicating significant academic impact. These included:

- Prajnya Ranganath [India], H-index 18 [12]
- Lenka Šlachtová [Czech Republic], H-index 17 [13]
- Güven Toksoy [Turkey], H-index 17 [14]

- Gemma Poke [New Zealand], H-index 16 [15]
- John Darlow [Ireland], H-index 15 [16]
- Eugenia Vicky Asare [Ghana], H-index 10 [17]

Conversely, some countries had profiles with lower H-indices, while others lacked clinical geneticists' profiles altogether, highlighting disparities in academic productivity across regions.

Conversely, some countries had clinical geneticists with lower H-indices (less than 9), including:

- André Travessa [Portugal], H-index 9 [18]
- Anita Barišić [Croatia], H-index 8 [19]
- Olivera Miljanovic [Montenegro], H-index 8 [20]
- Guillaume JOURET [Luxembourg], H-index 7 [21]
- Hind Alsharhan [Kuwait], H-index 7 [22]
- Tim Phetthong [Thailand], H-index 5 [23]
- Leniza De Castro-Hamoy [Philippines], H-index 4 [24]

Additionally, no clinical geneticist profiles were found for 155 countries (Afghanistan, Albania. Algeria, Andorra, Angola, Antigua & Barbuda, Argentina, Armenia, Austria, Azerbaijan, Bahamas, Bahrain, Bangladesh, Barbados, Belarus, Belize, Benin, Bhutan, Bolivia, Bosnia & Herzegovina, Botswana, Brunei, Brazil, Burkina Faso, Burundi, Cabo Verde, Cambodia, Cameroon, Central African republic, Chad, Chile, Colombia, Comoros, Congo democratic republic, Congo republic, Costa Rica, Ivory Coast , Cuba, Djibouti, Dominica, Dominican republic, East Timor, El Salvador, Equatorial Guinea, Eritrea, Estonia, Eswatini, Ethiopia, Fiji, Gabon, Gambia, Georgia, Grenada, Guatemala, Guinea, Guinea-Bissau, Guyana, Haiti, Honduras, Iceland, Indonesia, Jamaica, Jordan, Kazakhstan, Kenya, Kiribati , Kyrgyzstan, Laos, Latvia, Lebanon, Lesotho, Liberia, Libya, Liechtenstein, Lithuania , Madagascar, Malawi, Maldives, Mali, Malta, Marshall Islands, Mauritania, Mauritius, Mexico, Micronesia, Moldova, Monaco, Mongolia, Morocco, Mozambique, Myanmar, Namibia, Nauru, Nepal, Nicaragua, Niger, Nigeria, North Korea, Northern Ireland , Macedonia , Oman, Pakistan, Palau, Palestine, Panama, Papua New Guinea, Paraguay, Peru , Qatar, Romania, Rwanda, Saint Kitts & Nevis, Saint Lucia, Saint Vincent & the grenadines, Samoa, San Marino, São Tomé & Príncipe , Saudi Arabia, Scotland, Senegal, Serbia, Seychelles, Sierra Leone, Singapore, Slovakia, Slovenia, Solomon Islands, Somalia, South Africa, South Korea, South Sudan, Sudan, Sri Lanka, Suriname, Syria, Tajikistan, Tanzania, Togo, Tonga, Trinidad & Tobago, Tunisia, Turkmenistan, Tuvalu, Ukraine, United Arab Emirates, Uganda, Uruguay, Uzbekistan, Vanuatu, Venezuela, Vietnam, Wales ,Yemen, Zambia, Zimbabwe) , highlighting significant gaps in academic representation in the field of clinical genetics in various regions of the world.

Discussion

Clinical genetics encompasses a wide array of genetic disorders affecting individuals from diverse backgrounds [1, 2].

This study identified key figures in the developing world that have made substantial contributions to the field of clinical genetics. Notably, Aamir Jalal Al-Mosawi from Iraq stands out as one of the foremost pioneers in clinical genetics and dysmorphology in the developing world. His pioneering work in Iraq has significantly advanced the understanding and management of genetic disorders, especially in the areas of renal diseases, neurological conditions, inborn errors of metabolism, and rare genetic syndromes.

Aamir Jalal Al-Mosawi's work represents a cornerstone in the field of clinical genetics, particularly in the developing world. Al-Mosawi's groundbreaking contributions to the study of genetic disorders, rare syndromes, and therapeutic innovations have not only advanced the understanding of genetic diseases in Iraq but have also had global implications. Through his clinical practice and research, Al-Mosawi has expanded the horizons of genetic medicine, providing hope and improved outcomes for individuals with genetic disorders, both in Iraq and worldwide.

Aamir Jalal Al-Mosawi has been pioneering clinical genetics in Iraq contributing to advancing the knowledge about a variety of genetic disorders including renal disorders (Oculo-cerebro-renal syndrome, nephropathic cystinosis, nephronophthisis, adult polycystic kidney disease, polycystic disease of kidneys and liver , primary oxalosis, cystinuria, autosomal recessive steroid resistant syndrome, autosomal recessive proximal renal tubular acidosis, autosomal recessive distal renal tubular acidosis , X-linked dominant hypophosphatemic rickets , Laurence-Moon-Biedl syndrome, pediatric Bartter syndrome, congenital nephrotic syndrome (Finnish Type) [27-38], mental retardation [39], psychiatric (Gilles De La Tourette Syndrome and autosomal recessive autism) [40-42], skeletal (Achondroplasia and Ekman-Lobstein syndrome) [43-45], neurological (Semmola-Meryon-Duchenne syndrome, Rett syndrome and Huntington disease) [46-48], Inborn errors of metabolism such as Lesch-Nyhan disease, storage disease like Mucopolysaccharidoses and Niemann-Pick disease, [49-52], and hematological diseases such as Wiskott Aldrich Syndrome [53].

Dr. Al-Mosawi has made remarkable contributions to the study of chronic renal failure, renal tubular disorders, and nephropathic cystinosis among Iraqi children. His meticulous research and clinical observations have elucidated the patterns, clinical manifestations, and management strategies for these complex conditions, providing valuable insights into the spectrum of renal diseases prevalent in the Iraqi pediatric population [27-38]. His clinical work was not restricted to the commoner disorders (Thalassaemias, hemophilias, and Down syndrome), and the less common genetic disorders (Achondroplasia, polycystic kidney disease, Semmola-Meryon-Duchenne syndrome Werding Hoffman disease, Gaucher disease, Mucopolysaccharidoses, Wilson's disease) [25, 26, 54-59].

He was keen in documenting rare syndromes such Klinefelter syndrome associated with renal Fanconi syndrome, Diastrophic dysplasia, and Cornelia De Lange syndrome [60-62]. These studies not only expanded our knowledge base but also highlighted the complexities of diagnosis and management in rare genetic conditions.

Furthermore, he documented the occurrence of many of rare malformation syndromes such as Mostyn-Embrey syndrome [63, 64].

Dr. Al-Mosawi has been instrumental in documenting and providing clinical insights into many rare genetic and non-genetic disorders. His work has expanded our understanding of genetic variability and the challenges involved in diagnosing and managing these conditions.

Extremely rare medical disorders on the global level described by Iraqi pediatricians

Rare genetic disorders pose significant diagnostic challenges, often necessitating extensive medical consultations before a

conclusive diagnosis is reached. Aamir Jalal Al-Mosawi's case reports contribute valuable clinical insights into rare conditions, shedding light on their variability and expanding our understanding of genetic manifestations.

Aamir Jalal Al-Mosawi published at least thirty-one articles documenting very rare genetic and non-genetic disorders , including two cases of Coffin Siris syndrome, the thirty fifth and the thirty sixth reported cases of cutis laxa type II (Debre type), the first case of Aicardi syndrome in the Arab with the novel occurrence of corneal opacity, the case number 130 of Townes Brocks syndrome, the sixty fourth case of pediatric Churg Strauss syndrome, the second case of pediatric unilateral Vogt Koyanagi Harada syndrome in the world, the first case of Von Recklinghausen syndrome associated with right-sided basal ganglion abnormalities on brain magnetic resonance imaging, the twenty eighth case of congenital Chevalier Jackson syndrome, a case of Adams Oliver syndrome associated with evidence of periventricular calcifications on brain CT-scan, a case of the syndrome of congenital facial palsy and unilateral anotia, the third case of the extended Michelin tire baby syndrome associated with undescended testis, mental retardation and hearing impairment, two cases of Noonan syndrome; one associated with CT-scan evidence of brain atrophy and ventriculomegaly and one associated with unique multiple cardiac defects, the 58th Case of Toriello-Carey Syndrome, a case of Dandy walker syndrome with unusual presentation and unusual radiologic sign on CT-Scan, two cases of non-syndromal hemifacial microsomia, a case of Mowat Wilson syndrome associated with pseudo rocker bottom feet deformity, the thirty fourth and thirty fifth cases of Goldberg Shprintzen syndrome, the case number 52 of Ruprecht Majewski-Bosma syndrome, the case forty-one of crossed unfused renal ectopia, the second case Davis Buckley hyperimmunoglobulin E syndrome associated with IgA deficiency, the first reported case of posthitis in circumcised child, the case number 112 of congenital chylous ascites, the fourth case of familial non-syndromic gastric neuroendocrine-adenocarcinoma in the world, and the second case of Bardet-Biedle syndrome associated with keratoconus.

The 31 articles highlighted clinical presentations, diagnostic challenges, and implications for management. Many cases provided unique insights into genetic variability and many articles underscored the importance of meticulous clinical evaluation in achieving accurate diagnoses and tailored treatment strategies. Some case expanded the clinical spectrum, emphasizing the variability within syndromes and the implications for genetic counseling and therapeutic interventions [65-95].

New clinical conditions including new clinical disorders, new clinical syndromes and new clinical associations described by Aamir Jalal Al-Mosawi

Dr. Al-Mosawi's research has also led to the discovery and documentation of new clinical disorders, syndromes, and associations. These discoveries have broadened the clinical spectrum of these conditions and have profound implications for genetic counseling and treatment strategies.

Aamir Jalal Al-Mosawi reported fourteen documented new clinical conditions including new clinical disorders, new clinical syndromes and new clinical associations description including Idiopathic hyperuricosuria, hypercalciuria, and infantile renal stone disease [96], a dysmorphic girl with acrocephaly, seizures, long spindle fingers, and cherry red spots [97], a new congenital syndrome associated with mental-growth retardation, microphthalmia, microcornea, iris, and uvea colobomata, transient ocular hypopigmentation, contralateral optic disc colobomata, and dilated third ventricle [98], A new clinical disorder associated with hypoparathyroidism, vitiligo, poliosis, and macrocytic anemia [99], a new congenital syndrome characterized by facial dysmorphism, mental retardation, triphalangeal toes and unilateral renal agenesis [100], a new congenital syndrome characterized by unique facial dysmorphology consisting of reverse slanting of split eyebrows and palpebral fissures [101], A new dysmorphic syndrome characterized by psychomotor retardation, low set ears, retrognathia, facial dysmorphism, and schizencephaly [102], a new congenital syndrome characterized by congenital partial hemihypertrophy, low set ears, hypertelorism, and epicanthi folds [103], a new clinical disorder associated with mental retardation, periventricular white matter hyperintensity on brain MRI, retinitis pigmentosa, and optic atrophy [104], a new genetic disorder of autosomal dominant non-syndromic unilateral renal hypoplasia [105], A new psychiatric-endocrinological association consisting of atypical autism associated with elevated gonadotrophin and precocious puberty [106], a new genetic neurologic syndrome with characteristic neuroimaging findings, severe cerebral palsy associated with hydrocephalus, and mutation of Kinase D-Interacting Substrate of 220-kDa (Kidins220) gene [107], a new variant of childhood Lennox-Gastaut syndrome associated with low set ears and unilateral cryptorchidism [108], A new psychiatric-neurologic disorder associated with cerebral palsy, autism and periventricular white matter hyperintensity on brain MRI [109].

Dr. Al-Mosawi's investigations into therapeutic interventions for conditions like Huntington's disease of childhood and pediatric Bartter syndrome underscored the evolving landscape of genetic therapeutics. He delved into rare syndromes, therapeutic challenges, and diagnostic innovations, striving to enhance our understanding of genetic conditions and improve patient care.

The contribution of Aamir Jalal Al-Mosawi to diagnosis and treatment of international patients with genetic conditions

The first documented case of Niikawa-Kuroki syndrome in Kazakhstan was reported by Dr. Aamir Jalal Al-Mosawi and Fewin, demonstrating his involvement in diagnosing rare genetic disorders in international settings. The case underscored the importance of accurate diagnosis and multidisciplinary management in addressing rare syndromes [110].

Dr. Aamir Jalal Al-Mosawi's diagnostic acumen in familial Mediterranean fever (Sheppard Siegal syndrome) was illustrated in his report about a patient living in the United Arab Emirates. He emphasized in this paper, the role of serum amyloid A in guiding treatment decisions. This case highlighted the value of integrating clinical expertise with biomarker analysis in rare genetic conditions [111].

Dr. Al-Mosawi's contributed to the treatment of a patient with Williams syndrome from India, emphasizing the integration of evidence-based medicine with expert opinion to optimize patient outcomes [112].

This study highlights the disparities in academic productivity in clinical genetics across different regions, with several countries lacking significant representation in the field. This underscores the need for greater investment in clinical genetics education and

research, particularly in underrepresented regions.

This bibliometric analysis serves as a valuable resource for identifying key contributors to clinical genetics in the developing world, providing insight into the academic and clinical efforts that are shaping the future of genetic medicine in these regions.

Conclusion

This study underscores the significant contributions of clinical geneticists in the developing world, particularly individuals like Aamir Jalal Al-Mosawi, whose work has transformed the field of clinical genetics in Iraq and beyond. However, the study also highlights the academic disparities that persist in the field, emphasizing the need for increased investment in clinical genetics education, research, and infrastructure to ensure broader representation and progress across all regions of the world.

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The author has the copy right of all the sketches (Figures) included in this paper.

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