John Langdon Down was born in 1828 in an upstairs room above his father’s grocery shop in Torpoint. His father, Thomas Joseph Almond Down had failed in business on three occasions but he prospered in Torpoint. Torpoint was a working class village and the Down family lived among the artisans who worked in the naval base in Devonport. Langdon Down was taken out of school at the age of 14 and he spent the next four years behind the counter of his father’s shop. He had no higher education. At the age of 18 he had what might be described as a mystical experience. A heavy summer shower drove the family to take shelter in a cottage. “I was brought into contact with a feeble minded girl, who waited on our party and for whom the question haunted me - could nothing for her be done? I had then not entered on a medical student’s career but ever and anon... the remembrance of that hapless girl presented itself to me and I longed to do something for her kind." (J Down, 1879)

His first step on the road to a medical career was to go to London and to be apprenticed to a surgeon practitioner in the East End, where he learned the basic skills of blood letting, applying blisters, extracting teeth, and dispensing simple medicines. He quickly realised that without some knowledge of the sciences the medical qualifying examinations would prove to be too difficult for him.

One of the best basic science courses in London was offered by the Royal Pharmaceutical Society. He registered as a student and he quickly distinguished himself, taking the two professional examinations of the Society in one year. It was not his intention to practice as a pharmacist and he never registered as a member of the Pharmaceutical Society. He went back to Torpoint and with his newly acquired knowledge and skills he developed a very successful series of over the counter products which boosted the turnover of his father’s shop. Local directories described the enterprise as that of “grocers, chymysts, druggists, linen and woollen drapers.”

The Royal Pharmaceutical Society needed a new laboratory assistant. Langdon Down was headhunted and he went back to the laboratories of the Pharmaceutical Society where his main duty was to assist students with their bench work. This diversion was short lived. He became
ill, probably with tuberculosis, and he returned to Torpoint, where with rest, leisure, and fresh air he gradually recovered. His father died in 1853 and Langdon Down returned to the career of his choice. He entered the medical school of the London Hospital. He was a brilliant student and in his final year he took gold medals in medicine, surgery and obstetrics and the medal for the best student of the year.

Before leaving Torpoint he had written a prize essay on “Nature’s Balance” with the title “The Wisdom and Beneficence of the Creator, as displayed in the compensation between the animal the vegetable kingdoms.” He presented a copy to Mary Crellin. They were both very religious people. She read it, underlined it and kept it. The relationship flourished and they married in due course. When he took the qualifying examinations of the Royal College of Surgeons and of the Worshipful Society of Apothecaries he was appointed resident accoucheur. This gave him free board and lodgings and he combined his obstetric duties with further study, taking the London MB two years later, again with great distinction. The ink on his university parchment was scarcely dry when he was appointed Medical Superintendent of the Royal Earlswood Asylum for Idiots. He had no specific experience to equip him for the supervision of a very large institution for handicapped people of all ages. Earlswood had lost credibility in the years immediately preceding 1858. It had been criticised by the Commissioners in Lunacy and also in the press. A new broom was needed. John Conolly, the reformer of the psychiatric institutions was Visitor to Earlswood and he was the one to steer Langdon Down through the rough seas of his new venture.

John Conolly was an ardent ethnologist, one of a declining number. Ethnology attempted to correlate a relationship between the external contours of the skull and the degree of development of underlying areas of the brain. Each area of the brain was assigned a specific psychic or intellectual function. In parallel anthropology was in fashion and the classification of head shapes in different races was thought to correlate with different potential learning skills. Blumenbach’s Latin and German papers were translated into English by Bendyshe in 1865 (Bendyshe, 1865). Blumenbach’s classification of the races of the world into Mongolians, Aztecs, Caucasians, Malayans and Ethiopians was highly popular. Langdon Down read Bendyshe’s book and he immediately set about endeavouring to assign all the Earlswood residents to one or other of Blumenbach’s racial groups. This was how he came to publish his ethnic classification. The basis of his ethnic classification was the measurement of the diameters of the head and the identification of specific facial features from photographs which he took himself. Over 200 of his photographs have survived, making the collection the largest known archive of clinical photography of the Victorian era.

His ethnic classification never came to be widely accepted and indeed he himself abandoned it in due course. All that is now remembered of it is his description of what he described as the Mongolian type. He is sometimes criticised as a racist. His opening statement in which he sets out the predominance of Caucasian types is overlooked. He said: “of course there are numerous representatives of the great Caucasian family”. His description of the Mongolian group led to the specific recognition of these residents as a distinct category and in due course to the designation of those whom he had described as Mongolian as having Down’s syndrome. He published his paper in the London Hospital Reports (J Down, 1862) and he published it again verbatim in the Journal of Medical Science the following year. It was to be reprinted yet again in 1887, when, in association with the text of his three Lettsomian Lectures at the Medical Society of London he presented once again the papers which he had previously written on neurological subjects. (J Down, 1887). His papers frequently refer to idiots, and imbeciles. These were descriptive terms then commonly used but now abandoned by consent.

Of the Mongolian group he wrote: “the great Mongolian family has numerous representatives and it is to this division I wish, in this paper, to call special attention. A very large number of congenital idiots are typical Mongols. So marked is this that, when placed side by side, it is difficult to believe the specimens compared are not children of the same parents. The number of idiots who arrange themselves around the Mongolian type is so great, and they present such a close resemblance to one another in mental power, I shall describe an idiot member of this racial division, selected from the large number that have fallen under my observation.

The hair is not black, as in the real Mongol, but of a brownish colour, straight and scanty. The face is flat and broad, and destitute of prominence. The cheeks are roundish and the internal canthi more than normally extended laterally. The eyes are obliquely placed, and it is to this division I wish, in this paper, to call special attention. A very large number of congenital idiots are typical Mongols. So marked is this that, when placed side by side, it is difficult to believe the specimens compared are not children of the same parents. The number of idiots who arrange themselves around the Mongolian type is so great, and they present such a close resemblance to one another in mental power, I shall describe an idiot member of this racial division, selected from the large number that have fallen under my observation.
the levatores palpebrarum derive from the occipito-frontalis muscle in the opening of the eyes. The lips are large and thick with transverse fissures. The tongue is long, thick and much roughened. The nose is small. The skin has a slight dirty yellowish tinge, and is deficient in elasticity, giving the appearance of being too large for the body.

The boy’s aspect is such that it is difficult to realise that he is the child of Europeans, but so frequently are these characters presented that there can be no doubt that these ethnic features are the result of degeneration.

The Mongolian type of idioccy occurs in more than ten per cent of the cases which are presented to me. They are always congenital idiots and never result from accidents after uterine life. They are, for the most part instances of degeneracy arising from tuberculosis in the parents. They have considerable power of imitation, even bordering on being mimics. They are humorous and a lively sense of the ridiculous often colours their mimicry. This faculty of imitation can be cultivated to a very great extent and a practical direction given to the results obtained. They are usually able to speak; the speech is thick and indistinct, but may be improved very greatly by a well directed scheme of tongue gymnastics. The co-ordinating faculty is abnormal, but not so defective that it cannot be strengthened. By systemic training, considerable manipulative power may be obtained.

The circulation is feeble and however much advance is made intellectually in the summer, some amount of retrogression may be expected in the winter. Mental and physical capabilities are, in fact, directly as the temperature. The improvement which training affects in them is greatly in excess of what would be predicated if one did not know the characteristics of the type. The life expectancy, however, is far below the average, and the tendency is to the tuberculosis which I believe to be the hereditary origin of the degeneracy.

Apart from the practical bearing of this attempt at an ethnic classification, considerable philosophical interest attaches to it. The tendency in the present day is to reject the opinion that the various races are merely varieties of the human family having a common origin, and to insist that climatic, or other influences are insufficient to account for the different types of man. Here however we have examples of retrogression or, at all events of departure from one type and the assumption of the characteristics of another. If these great racial divisions are fixed and definite, how comes it that disease is able to break down the barrier, and to simulate so closely the features of the members of another division? I cannot but think that the observations which I have recorded are indications that the differences in the races are not specific but variable. These examples of the results of degeneracy among mankind, appear to me to furnish some arguments in favour of the unity of the human species."

His description covers the most important features of Down syndrome, with the exception of the flattening of the back of the head. He added this observation to his description in his first Lettsomian Lecture. He was undoubtedly aware of the skull shape. The admission examination in Earlswood included specific skull measurements. The practice of measuring the diameters of the head and probably been introduced by John Conolly as part of the examination required under the Lunacy Act which governed the operation of Earlswood. Langdon Down retained one typical skull vault for future demonstration. This is now in the museum of the Royal London Hospital. He also endeavoured to photograph the brain but the picture is not well focused.

His son Reginald made a further important observation. Reginald did not make any formal contributions to the medical literature but in 1909 in contributing to discussion of a paper by Shuttleworth he passed around hand prints of a number of patients with Down syndrome showing that “the bones of the palm differed from the normal in their extreme irregularity, and the tendency of the principal fold-lines to be two in number only, instead of three as was most commonly the case.” Reginald may have identified this peculiarity himself or his father may have shown it to him. A sketch of the palmar crease pattern dated 1908 survives in the family papers (R Down, 1909). Regrettably his philosophical views were not those of his father. Speaking at a meeting of the Medico-Psychological Society he said that the Mongolian features were accidental and superficial and that as there were other features which were in no way characteristic of the Mongolian race the abnormality "must be a reversion to a type even further back than the Mongol stock, from which some ethnologists believe all the various races of men have sprung.” (R Down, 1905) This statement was to be quoted later by Crookshank whose "A Mongol in our Midst" postulated that Mongolian imbecility as he described it, represented regression to the characteristics of the Orang Utan (Crookshank, 1924). His father
would have disapproved. Reginald's suggestion may have been to some degree related to his reaction to a personal problem. He was a disappointed father, his first son, born in 1905, having Down syndrome. At the time of Reginald's communication his son was aged three years old and the long term implications must have just then begun to become apparent to his father. Reginald's wife Jane never came to terms with their son's problems. The boy however grew to manhood in the Langdon Down home and he came to be a well-loved member of the family, living a happy life to the ripe old age of 65.

It took some time for Langdon Down's identification of the syndrome to make an impact. In 1876, ten years after Down's publication, Mitchell and Fraser published an account of what they described as Kalmuc idiocy, noting at the time that they had searched the medical literature and they had found no previous account of the condition. What they described was indeed Down's syndrome and they had failed to note his earlier publication in the same journal in which their paper later appeared. The first reference to Langdon Down's ethnic classification was probably in 1877 (Ireland). Two years later Tanner and Meadows also referred to it. (Tanner and Meadows, 1879). Shuttleworth in 1886 referred to Langdon Down's ethnic classification and he included the "Mongol type" in is tabulation. (Shuttleworth, 1886). Following on the publication of the Lettsomian Lectures references became more frequent, beginning with Goodheart in 1888 (Goodheart, 1888). In the United States Brush contributed a section to the Cyclopaedia of Diseases of Children in 1891 in which he quoted Down's description at length. (Brush, 1891). Subsequently other authors included the description in major publications, sometimes without attribution. (Tredgold, 1903).

By the turn of the century, Mongolism had become a widely used descriptive term. This was the title used by Bertram Hill in 1908 and by Penrose as late as 1961 (Hill, 1908; Penrose, 1961). The term was gradually dropped from 1961, to be superseded by Down syndrome but it was still used occasionally and the 1967 Ciba Symposium on the disorder chaired by Lord Russell Brain was entitled Mongolism. In 1961, a very prestigious group of genetic experts wrote a joint letter to the Lancet which read: "It has long been recognised that the terms "Mongolian Idiocy", "Mongolism", "Mongoloid", etc as applied to a specific type of mental deficiency have misleading connotations. The importance of this anomaly among Europeans and their descendants is not related to the segregation of genes derived from Asians; its appearance among members of Asian populations suggests such ambiguous designations as "Mongol Mongoloid"; increasing participation of Chinese and Japanese in investigation of the condition imposes on them the use of an embarrassing term. We urge, therefore, that the expressions which imply a racial aspect of the condition be no longer used. Some of the undersigned are inclined to replace the term Mongolism by such designations as "Langdon Down Anomaly", or "Down’s Syndrome or Anomaly", or “Congenital Acromicria”. Several of us believe that this is an appropriate time to introduce the term “Trisomy 21 Anomaly”, which would include cases of simple Trisomy as well as translocations. It is hoped that agreement on a specific phrase will soon crystallise once the term “Mongolism” has been abandoned.” (Allen et al, 1961).

The first two signatories were Allen and Bender, the former a world authority on mental retardation and the latter the author of a book on Mongolism. Allen had initiated the redesignation project in Bethesda in Maryland. The signatories represented the cream of the investigators who were active in the burgeoning field of genetic studies. The UK signatories were Carter, Ford, Penrose, Polani and Langdon Down. According to Polani, Penrose approached Norman Langdon Down to ask for the permission of the family in proposing the new descriptive term. Norman was a grandson of John Langdon Down. As a matter of interest the Lancet gave him the wrong initials designating him as W Langdon Down. He was the medical superintendent of Normansfield, and this responsibility had passed from father to sons to grandson for a period of 102 years. Normansfield had been opened by Langdon Down in 1868 to cater for the upper classes. There were only two objections to the proposals and in due course the Editor of the Lancet issued his ruling: “Down’s Syndrome is an appropriate alternative for Mongoloid Idiocy until the chromosome abnormality in the disorder has been fully elucidated and a new scientific term has been coined.” The redesignation of the condition was confirmed by the World Health Organisation in 1965. The People’s Public of Mongolia had approached the Director General and said that they objected to the use of the descriptive term "Mongolian Idiot" as it was derogatory to them. Down’s Syndrome was adopted as an official definition. This decision has never been revoked. (Beighton & Beighton, 1986).

John Langdon Down was an astute clinical observer. He was the first to describe Prader-Willi Syndrome (Ward, 1997). He contributed
the only autopsy report available to Little when he described the relationship between perinatal crises and subsequent cerebral palsy. (Little, 1862) He carried out the autopsy which makes it possible to say that the index case of West’s syndrome was cryptogenic (Ward, in press) He wrote papers on pseudohypertrophic muscular dystrophy, microcephaly and plagiocephaly.

Langdon Down published an almost-forgotten book on the “Education and Training of the Feeble in Mind.” (Down, 1876). He advocated what he described as a medical model of management, but in effect the specifically medical content of the programme was very limited. He did however, stress the importance of diet although the scientific basis of dietetics was as yet not established. In the Royal Earlswood Asylum, and later in Normansfield, he set up training systems based on physical exercise, sensory stimulation, and role-playing in such social activities as shopping. This involved training and encouraging a team of carers who would now be classified as play therapists, occupational therapists, speech therapists and specialist teachers. None of these groups had as yet emerged as distinct professions. He recognised that social exclusion and the loneliness of limited social contact were major problems for all classes of society. Among the well-to-do children with handicaps spent their days isolated in the servants’ quarters of their homes. Those in the middle classes were neglected in school and were perceived as a poor educational investment. Those in the lower income groups placed an impossible burden on their struggling parents. He recommended institutional training as it was only in institutions that the range of necessary services would be provided. He hoped that many could later be integrated in useful activity at home. He also emphasised that by putting children of comparable talents in groups together they could learn more easily, protected from the pressure of competition. In the conditions of the time, the social attitudes, the transport problems, and the overall lack of services his ideas were practical and innovatory. In particular, although his own Normansfield centre was orientated towards the upper classes, he laid great emphasis on the importance of the provision of services for lower income families funded either by single counties or by groups of counties. In the Royal Earlswood Asylum, the charitable institution which he served for ten years, his sweeping reforms had established for it a world-wide reputation as a centre of excellence.

Aside from his contribution to medicine he was in advance of his time in being prepared to accept the advancement of women in medicine, the law and the church. His Harley Street consulting rooms were used for fundraising for the suffragette movement. When he died shops closed and members of the public stood on the pavement in silent tribute as his cortège passed by. A street was named in his honour in Teddington and another in Torpoint. Surprisingly his life story has not attracted the attention it deserves. More than one hundred years after his death Langdon Down’s contributions to medicine have been commemorated at the Mansell Symposium in the Medical Society of London (Ward, in press) and a biography has been published by the Royal Society of Medicine (Ward, 1998).

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