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HISTORICAL & PERSONAL PERSPECTIVES

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Julia Bell and the *Treasury of Human Inheritance*

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Abstract The *Treasury of Human Inheritance* represents the most extensive, and one of the earliest series of documentations and analyses of human genetic disorders. Published between 1909 and 1958, from The Galton Laboratory, London, most of the numerous sections were written by Julia Bell, who represents a key figure in the development of human and medical genetics. Her combination of mathematical training, genetic knowledge and clinical expertise yielded numerous important insights into human inheritance first appearing in the *Treasury*; it remains a valuable scientific as well as an historical record of the genetics of a range of important inherited disorders.

Introduction: the *Treasury of Human Inheritance*

Among the works linking human genetics with the study of inherited disorders, the *Treasury of Human Inheritance* stands as a unique record for its era. Spanning half a century from its initial volume in 1909, it provides both a compilation and an analysis of knowledge at the time about many of the key disorders that have provided much of the foundation for our subsequent understanding from clinical and molecular genetic studies. Yet, despite this and the fact that it remains in print and available after almost a century, it is now little known, as is the work and life of the remarkable physician and scientist, Julia Bell, who was the core of the *Treasury* and provided most of the original findings arising from it, as well as the painstaking and meticulous documentation that it is based upon.

This article aims to make the importance of the *Treasury of Human Inheritance* more widely known and valued to clinical workers and scientists in human and medical genetics. It does not attempt to give a detailed analysis of its content, nor to trace the history of the individual disorders that it documents, something that can be found in the specific volumes of the work itself. The overall structure of the series is summarised in Tables 1 and 2.

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Julia Bell (1879–1979)

It is rare to find a single worker so completely identified with a major body of work, such as is represented by the *Treasury*, so a short summary of her life and career is given here. A fuller account, both original and informative, has been given by Bundey (1996), from which I have drawn. Figure 1 is reproduced from her paper.

With a life of a whole century spanning virtually the entire history of genetics so far, Julia Bell can be regarded as one of the key founders of both human and medical genetics. She had already been at Cambridge for 2 years (studying mathematics) when Mendelism was rediscovered in 1900, and she lived to see the first isolation of a gene. Debarred as a woman from taking a Cambridge degree, she received it, as did many women at the time, from Trinity College, Dublin. Her links to genetics began in 1908, when she began work for Karl Pearson at University College London, at first working from home, then at the newly founded Galton Laboratory as a statistician, being already present when he initiated the *Treasury of Human Inheritance* in 1909; her name, though acknowledged, is not associated with any of the specific parts of the initial volume.

It soon became clear that undertaking detailed family studies (and being accepted by clinicians) required a medical degree, so when publication of the *Treasury* was suspended at the outbreak of World War I, she enrolled at the age of 35 to study medicine at the Royal Free Hospital for women, the only medical school in England accepting women at that time. Returning to the Galton Laboratory in 1920, as a Research Fellow funded by the Medical Research Council, she was responsible for the

Table 1 *Treasury of Human Inheritance*: outline structure

Volume	Date	Parts	Editor	Authors
I	1909–1912	1–7	Karl Pearson	Various
II	1922–1932	1–5	Karl Pearson	Julia Bell
III	1925	1	Karl Pearson	P. Stocks
IV	1934–1947	1–5	R.A. Fisher/Lionel Penrose	Julia Bell
V	1951–1958	1–3	Lionel Penrose	Julia Bell

entire series of subsequent studies that made up volumes II, IV and V of the *Treasury*, no fewer than 13 essentially separate monographs, appearing at intervals of 2–3 years and making up much, though not all of her life's work.

During this period, she worked successively under the first three Galton Professors (Fig. 2)—Karl Pearson, Ronald Fisher and Lionel Penrose, though her work seems to have been almost entirely independent. She also collaborated closely with J.B.S. Haldane, notably on genetic linkage in haemophilia, and so was an integral part of the remarkable concentration of talent in genetics based around University College London during the early and middle decades of the 20th century.

Just as important was her role as an interface between basic geneticists and clinicians, especially ophthalmologists based at Moorfields Eye Hospital, such as Edward Nettleship, and neurologists at National Hospital for Nervous Diseases, Queen Square, London; these two specialities forming the subjects for the different parts of volumes II and IV, respectively, of the *Treasury*. Given the attitudes of those times in medicine towards women and towards non-clinicians, it would have been highly unlikely that she would have been accepted and respected (as she clearly was) as a collaborator, by the foremost clinicians of the time in Britain, without detailed medical knowledge as well as a medical qualification. Her

Table 2 *Treasury of Human Inheritance*: parts authored by Julia Bell (modern names for disorders given in *square brackets* where significantly different)

Volume	Part	Date	Title
II			Anomalies and diseases of the eye
	I	1922	Retinitis pigmentosa and allied disorders Congenital stationary night-blindness. Glioma retinae [retinoblastoma]
	II	1926	Colour-blindness
	III	1928	Blue sclerotics and fragility of bone [osteogenesis imperfecta]
	IV	1931	Hereditary optic atrophy (Leber's disease).
	V	1932	On some hereditary structural anomalies of the eye and on the inheritance of glaucoma.
IV			Nervous diseases and muscular dystrophies
	I	1934	Huntington's chorea
	II	1935	On the peroneal type of progressive muscular atrophy [Charcot-Marie-Tooth disease].
	III	1939	On hereditary ataxia and spastic paraplegia.
	IV	1943	On pseudohypertrophic and allied types of progressive muscular dystrophy.
	V	1947	Dystrophia myotonica [myotonic dystrophy] and allied diseases
V			On hereditary digital anomalies
	I	1951	On brachydactyly and symphalangism
	II	1953	On syndactyly and its association with polydactyly
	III	1958	The Laurence–Moon syndrome.

Fig. 1 Julia Bell as a graduate and in later life. Reproduced from Bundey (1996) by permission of Journal of Medical Biography

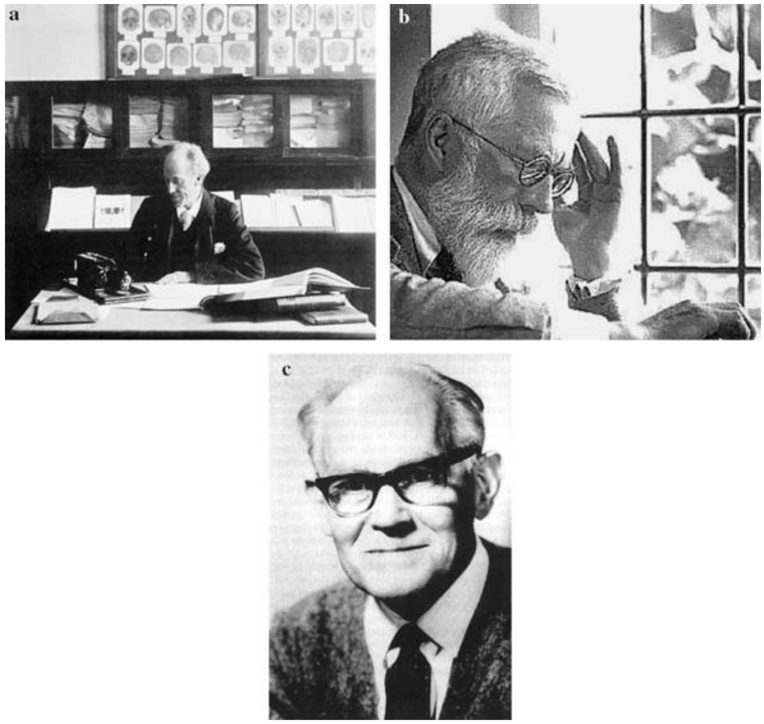

Fig. 2 The three Galton Professors responsible for the *Treasury of Human Inheritance*. **a** Karl Pearson; **b** Ronald Fisher (courtesy of Fisher Memorial Trust, copyright A. Barrington Brown); **c** Lionel Penrose (courtesy of Dr. Shirley Hodgson)

clinical skills can be judged not only by the documentation of cases in the different volumes, but by her having passed the examinations of the Royal College of Physicians of London, being elected Fellow in 1938.

Julia Bell's long career did not end with her formal retirement from the Galton Laboratory; having outlived all three Galton Professors, she continued work in association with the Laboratory and with the National Hospital, Queen Square until the age of 85 and remained actively interested until over 90; Bundey quotes her at almost 90 stating: 'I recommend old age—it can be full of interest'. Living independently until the age of 96, she reached the age of 100 years, the only centenarian I can find a record of in the sphere of human genetics.

The *Treasury of Human Inheritance*: sections prior to Julia Bell

The eight parts of volume I, appearing at intervals between 1909 and 1912 (Fig. 3) and some being very brief, contain much interesting information, with careful documentation of clinical data, pedigrees and previous literature. They represent a compilation, however, not an analysis, and tell us almost nothing about inheritance. The main topics covered included diabetes insipidus, polydactyly and brachydactyly, 'deaf mutism', congenital cataract and haemophilia, but also some more diffuse categories such as 'ability' and 'insanity'. Among the information most relevant to modern workers is the list of anomalies associated with congenital deafness ('deaf mutism'), the documentation of fetal abnormalities in cleft-lip and palate and the numerous photographs (including one of a hare as a comparison for 'hare lip'!). Dwarfism (parts VII and VIII) is fully documented by clinical photographs, X-rays and skeletal preparations. The part on haemophilia (1911) clearly shows different transmission by the sexes, but there is no discussion on its inheritance (it should be remembered that X-linkage was not clearly documented at this time, while Pearson as editor was fiercely opposed to even the idea of Mendelism).

In fairness, it should be recognised that a compilation was precisely what Pearson intended. In the preface to each part he states:

> For a publication of this kind to be successful at the present time, it should.....be entirely free from controversial matter. The *Treasury of Human Inheritance* therefore contains no reference to theoretical opinions

By controversy, he probably meant that between the Biometricians and the Mendelians (Magnello 2004).

He also states, entirely reasonably, that

> students of heredity find great difficulty in obtaining easy access to material bearing on human inheritance. The published material is voluminous, scattered over a wide and often very inaccessible journalistic area. The already collected although unpublished material is probably as copious, but no central organ for its rapid publication in a standardised form exists at present.

It is perhaps ironic that the comprehensive collections of clinical material on inherited disorders represented by the *Treasury* were to become one of the key foundations

UNIVERSITY OF LONDON

FRANCIS GALTON LABORATORY FOR NATIONAL EUGENICS

TREASURY OF HUMAN INHERITANCE

EDITED BY

KARL PEARSON, F.R.S.

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VOLUME I

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The power of man in varying the future human stock vests a great responsibility in each fresh generation, which has not yet been recognised at its just importance, nor deliberately employed.

FRANCIS GALTON, *Inquiries into Human Faculty*, 1883.

LONDON:

PUBLISHED BY DULAU AND CO., LTD., 37, SOHO SQUARE, W.

1912

Fig. 3 Title page of the initial volume of *Treasury of Human Inheritance* for Mendelism, having been initiated with the objective of disproving its importance.

At this point, a word needs to be said about the relationship between the *Treasury* and the subject of Eugenics, at this time a field of considerable popularity internationally. Despite Pearson's insistence on objectivity, reflected in both the content and tone of the early parts of the *Treasury*, the connection was inevitably a close one. The whole initiative was organised (and funded) by Francis Galton, father of eugenics; Karl Pearson's Chair was in 'Eugenics' and the cover of these early sections of the *Treasury* gives its provenance as 'The Francis Galton Laboratory for National Eugenics'. It is thus perhaps surprising (and creditable) that the work should be as scientific and as free from eugenic overtones as indeed it was; in this respect it makes a striking contrast to some other work at the time, notably that of Davenport and colleagues based at the Cold Spring Harbor Laboratory in America (Davenport 1910).

In 1911, shortly before the conclusion of volume I of the *Treasury*, Francis Galton died, and Karl Pearson, who would later write his biography, issued a tribute as a supplement to volume I, including the extraordinary plate of portraits reproduced here (Fig. 4) which purports to show the origins of Galton's talent! In fact, the whole of volume I shows that the work had not fully detached itself from its less than scientific eugenic surroundings.

With the outbreak of World War I in 1914, publication of the *Treasury* was suspended. In the preface to the first part of volume II, to be devoted to hereditary eye disorders and anomalies, Pearson notes that its publication was scheduled for 1911, but it was 1922 before it actually appeared. The whole series of sections was called the *Nettleship Memorial Volume*, since Edward Nettleship, who had provided much of the material, had died in the intervening time. A valuable account of Nettleship's key role in interacting with both Karl Pearson and Bateson has been provided by Rushton (2000) and it is clear from correspondence between them that he also played a major role in developing the concepts of inheritance as well as gaining a sound foundation of evidence.

Julia Bell was involved in collecting material on families with inherited disorders from the earliest years, but her medical training between 1914 and 1920 gave her greater authority to develop the work and the Medical Research Council is acknowledged as supporting it from 1920. For the next 40 years it was to bear her stamp and progressively to develop the key features that make it of lasting scientific value.

Publication and funding of the *Treasury*

Despite the personal funding and endowments by Francis Galton of the various initiatives associated with his name, it seems that, at least after Galton's death, the *Treasury* was expected to pay its way. It was issued by subscription, probably no more than 200–300 copies, since Pearson states in the preface to volume III that a subscription issue of 500 would allow a reduction in price to two thirds or even one half of that actually charged.

Volume I was issued under the imprint of the London publisher Dulau, but thereafter Cambridge University Press became the publisher. Remarkably, almost 100 years after its initiation, almost all sections remain in print and available—surely a publishing record unique in itself for a scientific work of this nature! (Those interested in obtaining one of the limited numbers of remaining sets should contact Professor Sue Povey at the Galton Laboratory, Wolfson House, Stevenson Way, London.)

The *Treasury of Human Inheritance* under Julia Bell

These volumes, in particular volume II devoted to ophthalmological genetics (in five parts) and even more volume IV on neurological genetics (also in five parts), were all written by Julia Bell, and stand as the foundations on which these two major fields of human and medical genetics rest today. It is striking that the topics of the various parts (see Table 2) address the same disorders that involve the practising clinical geneticist today, and also those neurologists and ophthalmologists involved in genetics. These are also the areas in which the past two decades have shown remarkable developments in our understanding of the molecular pathology.

It is impossible to detail all the important insights that these volumes record, but Table 3 summarises some of the principal ones, a remarkable achievement for a single worker, even allowing for the fact that Julia Bell's analyses were largely based on the collected data of others (mainly clinicians). In general, the neurological parts (volume IV) contain more detailed genetic analyses and interpretations than do the earlier ophthalmological ones, but they all share a number of important features. These include a scholarly and detailed historical introduction; statistical analysis of all available data (notably ages of onset and death, correlation of ages of onset between sibs and parent and offspring, incidence and geographical distribution); a general account of clinical features and pathology (with details of individual cases in the appendices); full pedigrees; and copious references to the literature.

Of especial importance is the tabulation of full raw data, broken down usually by sex and parental transmission, in addition to the overall analyses, which give standard deviations where appropriate. This allows the later worker to go back to the tables and reanalyse the data to extract information of which Julia Bell was herself unaware. A few examples only can be given here, but they will give an idea of what current workers on any of the disorders covered might find of relevance.

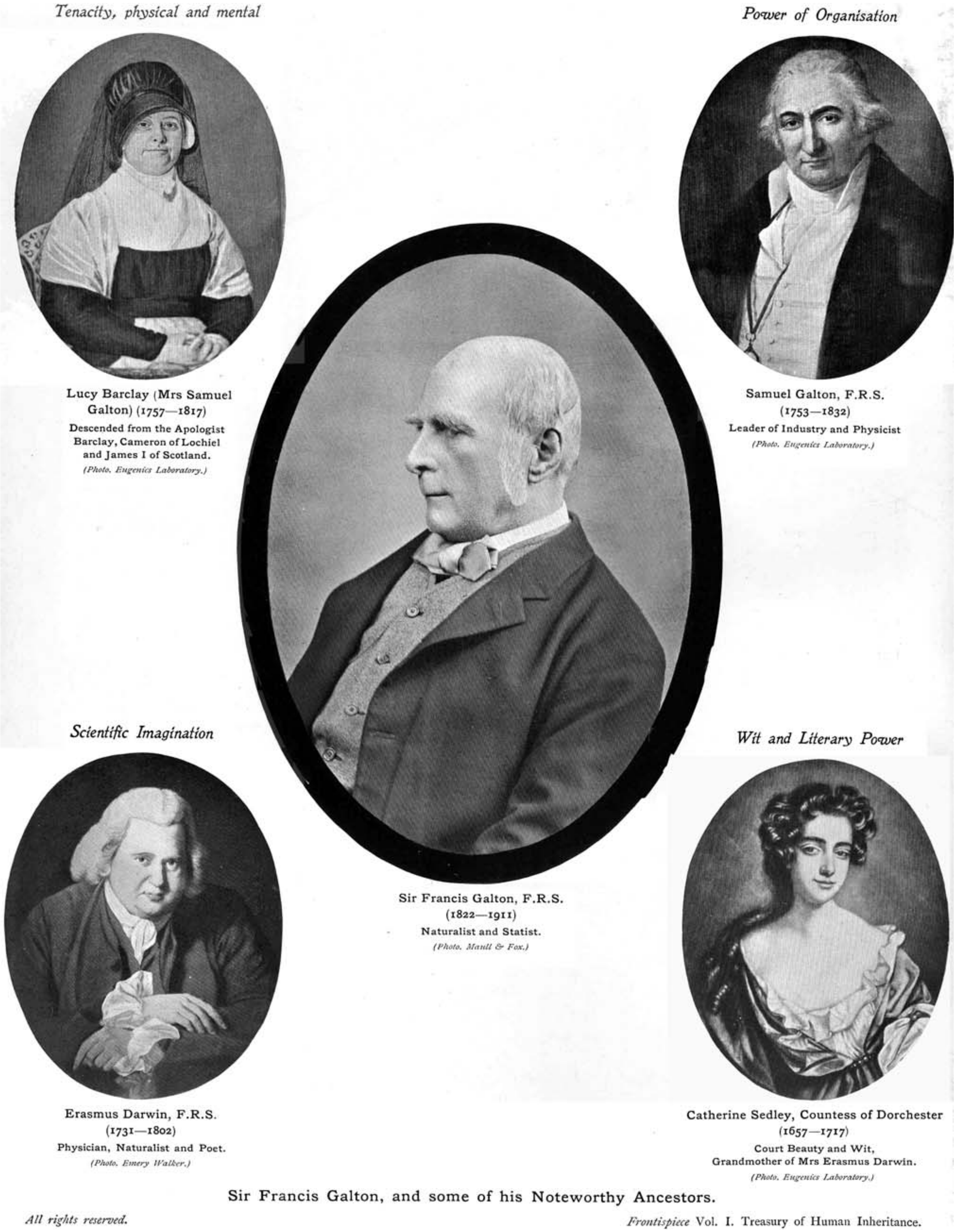

Fig. 4 Francis Galton and his ancestors. (Frontispiece to volume I of the *Treasury of Human Inheritance*)

The neurological sections making up volume IV show some differences in overall approach to the earlier volume II dealing with eye disease. The tabular material,

Table 3 *Treasury of Human Inheritance*: summary of major original findings from different volumes

Volume/part	Disorder	Finding
II. I	Retinitis pigmentosa	Specific associations with deafness (Usher syndrome) polydactyly (Bardet Biedl syndrome)
II. IV	Hereditary optic atrophy (Leber)	Overwhelmingly female transmission (95%)
IV. I	Huntington's disease	Quantitative analysis of age at onset, death; fertility, transmission. Possibility of presymptomatic detection predicted
IV. II	Peroneal muscular atrophy (Charcot-Marie-Tooth disease)	Recognition of genetic heterogeneity, notably X-linked form.
IV. III	Hereditary ataxia and spastic paraplegia	Autosomal recessive forms much earlier in onset than dominant forms. Clinical and genetic distinctiveness of Friedreich's Ataxia.
IV. IV	Pseudohypertrophic and allied forms of muscular dystrophy	Genetic classification used. Consanguinity only in autosomal recessive families. Late onset X-linked families recognised.
IV. V	Dystrophia myotonica	Analysis of anticipation. Recognition of childhood onset. Preferential male transmission in older generation. Distinction of myotonic dystrophy and myotonia congenita.
V. I	Brachydactyly and symphalangism	Classification of brachydactyly

graphs and statistical analyses are more extensive and detailed, the interpretation within a framework of Mendelian inheritance is more explicit, and there is a boldness and maturity in the discussion sections that reflect Julia Bell's own experience and confidence. The fact that Karl Pearson had been succeeded as editor by Fisher (and ultimately by Penrose) may also be significant. Nowhere is this more apparent than in the final part of volume IV (*Dystrophia myotonica and allied disorders*) where, despite utilising (and gracefully acknowledging) the extensive family material supplied by the London neurologist Maas, she firmly and explicitly rejects his conclusions that almost all family members showed minor signs of the disorder and that myotonic dystrophy and myotonia congenita were the same disorder.

Modern insights from the *Treasury*

Taking these chronologically, as summarised in Table 3, a few insights that have struck the author as particularly relevant to our modern understanding of inherited disease include the following:

Leber's optic atrophy (1931) Julia Bell recognised the almost exclusively maternal transmission of this disorder (95% in European families) and that this applied also to Japanese families where the actual sex ratio was predominantly female. She distinguished the inheritance from that of colour-blindness by lack of occurrence in the grandsons of affected males, but did not speculate on the possibility of cytoplasmic inheritance, which was left to Imai and Moriwaki in Japan (Imai and Moriwaki 1936). She rightly attributed the few families with male transmission to heterogeneity.

Huntington's disease (1934) This monograph can be considered as the foundation for many later studies, for its thorough analysis of age at onset and death (see Fig. 5), duration of disease, comparison of these against psychiatric or neurological features, fertility (unimpaired) and dominant inheritance pattern. It is also noteworthy for its prediction that genetics could be used for presymptomatic detection.

> The almost continuous anxiety of unaffected members of these families over so long a period must be a great strain and handicap, even if they remain free from disquieting symptoms; it is thus of urgent importance that some means should be sought by which the immunity of an individual could be predicted early in life, both from the point of view of relief to those who carry no liability to the disease and as an indication to others that they should abstain from parenthood. No facts in the clinical histories of patients provide definite guidance in this matter prior to the onset of symptoms, but the development of the science of genetics may at some future date enable us to obtain information concerning the inherent characteristics in such cases.

That linked genetic markers were meant by this statement is shown by the more specific statement 3 years later in her paper with J.B.S. Haldane (Bell and Haldane 1937) on genetic linkage between colour-blindness and haemophilia.

> The present case has no prognostic application, since haemophilia can be detected before colour-blindness. If, however, to take a possible example, an equally close linkage were found between the genes determining blood group membership and that determining Huntington's chorea, we should be able, in many cases, to predict which children of an affected person would develop this disease, and to advise on the desirability or otherwise of their marriage.

Edwards (2004) has shown how the general idea of using linkage for prediction was circulating in the writ-

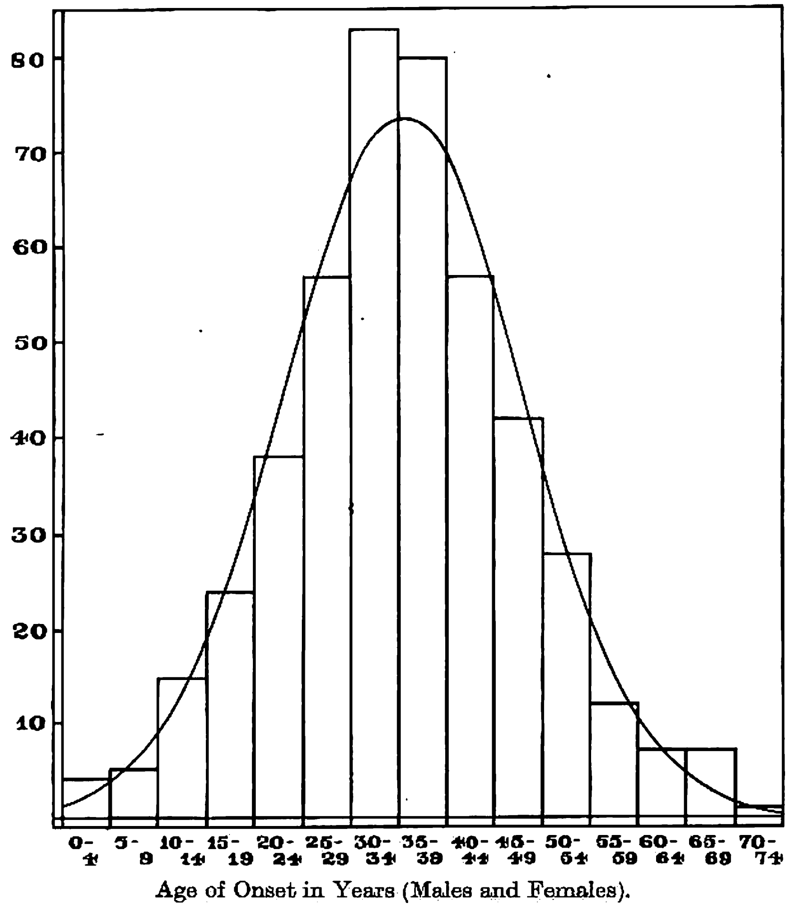

Fig. 5 *Graph* showing age at onset in Huntington's disease. (From *Treasury of Human Inheritance*, volume IV, part 1, p12)

ings of Haldane, Hogben and Fisher throughout the 1920s and 1930s, but it would be 50 years before DNA markers would make this a reality and open up a new era for the presymptomatic detection of late onset genetic disorders.

Charcot-Marie-Tooth disease (1935) Not only was the extensive heterogeneity recognised, but the X-linked form was clearly documented based largely on the unpublished observations of Findlay on a large Aberdeenshire kindred (Fig. 6), itself the foundation for subsequent molecular analysis (Haites et al. 1989). She also noted the likelihood that X-linkage might be responsible for a significant proportion of isolated male cases.

Hereditary ataxia and spastic paraplegia (1939) The very fact of bringing these conditions together in the same monograph indicates the modern classification, detailed in the text, of relating them to each other, while she makes clear the distinctive clinical and genetic features of Friedreich's ataxia.

X-linked muscular dystrophy (1943) Julia Bell's contribution here has been under-estimated (Emery and Emery 1995); not only did she estimate the proportions following different inheritance patterns (46% X-linked, 28% recessive, 26% dominant), but she found around 8% of X-linked families with onset at 15 years or later (these would now be considered the 'Becker' form), giving a graph suggesting that age at onset was continuous, not bimodal.

Myotonic dystrophy (1947) The author's own experience with this disorder has made him aware of how many aspects regarded much later on as 'new' findings, are in fact clearly recognised in Julia Bell's study. Thus, while the first clear recognition of the congenital form was by Vanier (1960), Bell's data on age at onset (Table 6 in the monograph) show onset under 10 years in 20% (42/218) overall, with onset under 5 years in 8% (17/218).

The data are broken down into 'parental' and 'offspring' generations, which show the proportion with onset under 10 years to be 25% (32/121) in the 'offspring' group, while for those with onset under 5 years, the proportion is around 12% (15/1121), proportions not dissimilar to those now recognised for childhood onset and congenital myotonic dystrophy. In discussing causes of death, she specifically noted that those dying under 5 years of age may be 'a special group with particular underlying causes'.

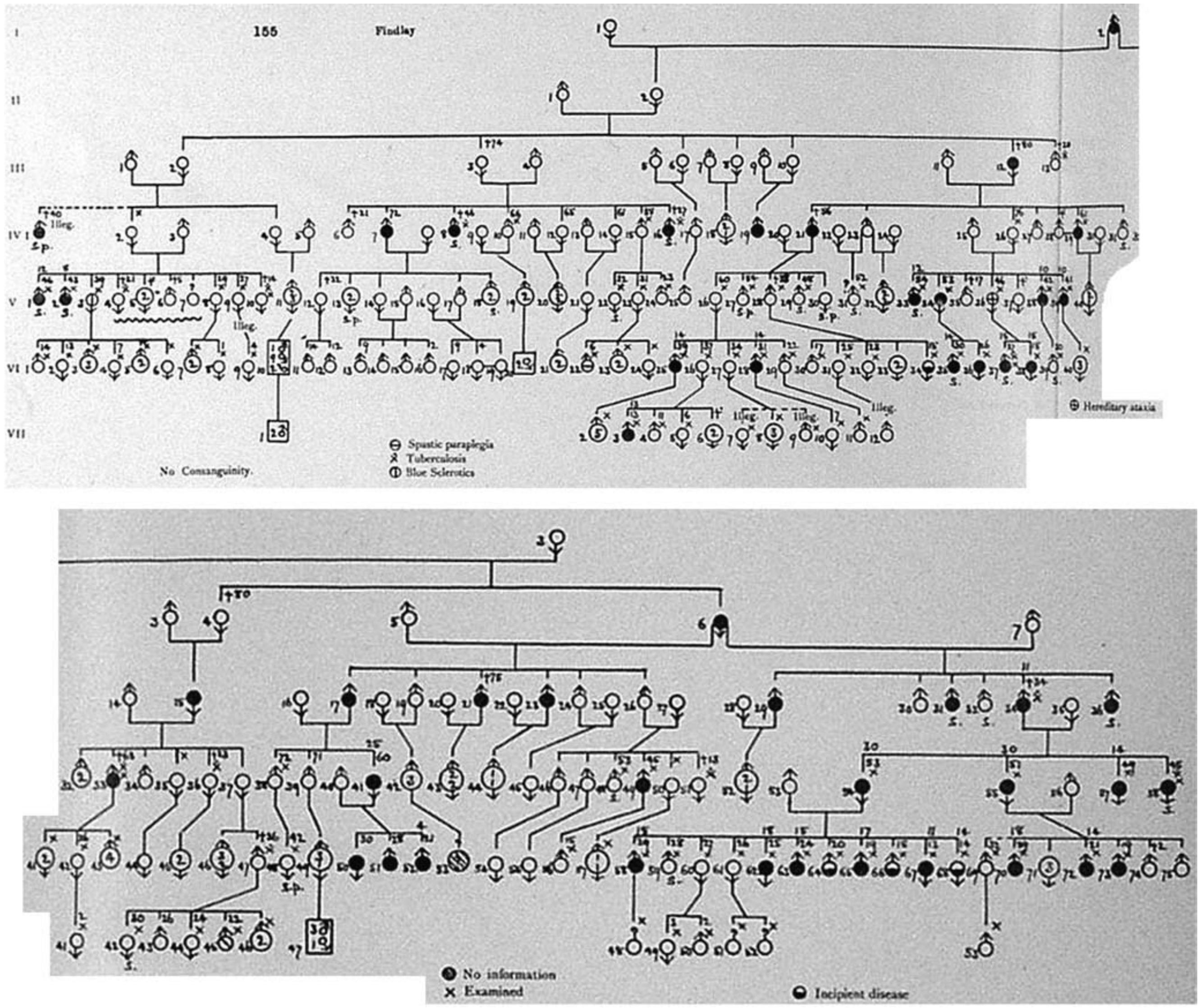

Fig. 6 Pedigree of X-linked Charcot-Marie-Tooth disease. (From *Treasury of Human Inheritance*, volume IV, part 2)

Another genetic aspect clearly identified in the study was the preferential paternal transmission, especially in the older generation with cataract only, or characterised as obligatory gene carriers without known clinical abnormality (Table 16 of the monograph). Overall paternal transmission was twice that of maternal, while for the cataract only/carrier group it was over three times as frequent. This finding was 'discovered' 40–50 years later by the author (Harper 2001) and Brunner et al.(1993), neither of us realising at the time that the data were plainly documented in Julia Bell's study! The explanation in terms of a greater degree of CTG repeat expansion in male meiosis is now at last clear (Harley et al. 1992).

Related to the above observations is the clear documentation on anticipation (called by Bell 'antedating') in the study (Table 6 in the monograph and Fig. 7 reproduced here). The statistical validity for earlier onset in the younger generation was clear ($\chi^2 = 47.558$), but her support for antedating is given somewhat obliquely in the text.

> There has been much conviction expressed from time to time that dystrophia myotonica provides a striking and undoubted example of antedating from one generation to the next; indeed, difficult as it always is to establish the fact of any inherent modification in the manifestations of a disease from one generation to the next, figures can be provided which certainly suggest that for one reason or another it is not possible here to assert the contrary.

It is quite possible that this caution may have reflected the views of Penrose (by now Galton Professor and editor of the *Treasury*), who a year later published an analysis of anticipation based on Bell's data (Penrose 1948) and concluded that it was the result of inherent variability of the disorder, biases of ascertainment and a possible allelic modifier. In this last factor he was strongly influenced by Bell's finding that the

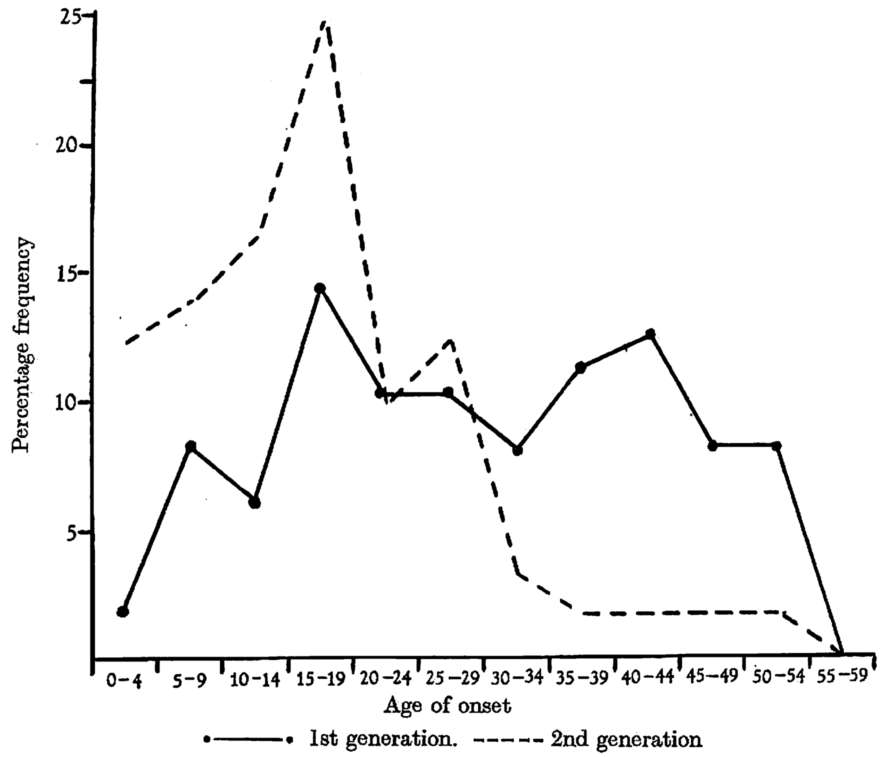

Fig. 7 Graph showing age at onset of myotonic dystrophy. (From *Treasury of Human Inheritance*, volume IV, part 5, p353)

parent–offspring correlation coefficient for age at onset (0.322) was much less than that between sibs (0.659), a difference not seen for other dominantly inherited disorders. It was not until the work of Höweler et al. (1989), closely followed by identification of the DNA instability characterizing the underlying mutation, that anticipation was fully validated and biologically explained (Harper et al. 1992).

The *Treasury of Human Inheritance*—some personal touches

While Julia Bell kept the text and analyses in the *Treasury* objective and restrained, she allowed herself some scope in the historical background and dedications for expressing her wider scholarly approach and her imagination. Thus, in the historical definition of 'ataxia' she provides the example, which may surprise current workers in the ataxia field:

> Byfield, in 1615, writing of angels, says:
> "we are not to thinke there is any ataxie among these glorious creatures."

Most parts have a quotation to occupy the otherwise blank space of the opening page, often with a photograph of the relevant key worker (e.g. Leber, Fig. 8). The quotations for the part on Huntington's disease strike a sombre note:

> "Twas slow: - Disease, augmenting year by year,
> Show'd the grim King by gradual steps brought near."
>
> Crabbe: *The Parish Register*
>
> "They do not live, but linger."
>
> Burton: *Anatomy of Melancholy*

Especially intriguing is the Chinese poem that introduces the volume on myotonic dystrophy; it is quite

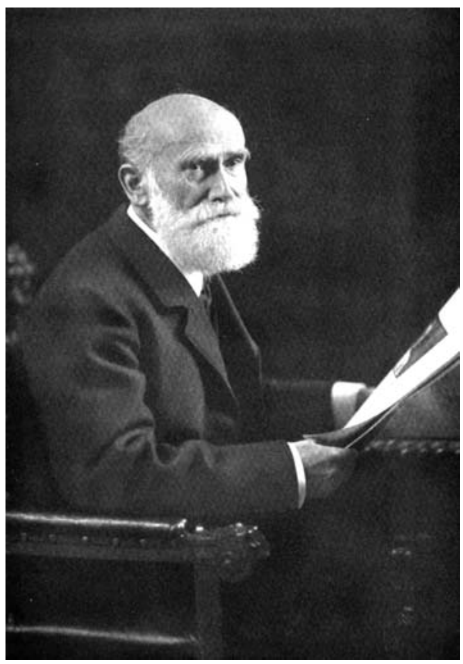

Fig. 8 Photograph of Theodor Leber. From *Treasury of Human Inheritance*, volume II, part 4 (frontispiece)

possible that Julia Bell knew the translator, Arthur Waley, who worked at the British Museum a short distance from the Galton laboratory.

Buddha's pity

Because he saw Mankind drowning in the Great Sea of Birth, Death and Sorrow, and longed to save them
For this he was moved to pity.
Because he saw them living in an evil time, subjected to tyrannous kings And suffering many ills, yet heedlessly following after pleasure
For this he was moved to pity.
Because he saw them living in a time of wars, killing and wounding one
Another, and knew that for the riotous hatred that had flourished in their
Hearts they were doomed to pay an endless retribution
For this he was moved to pity.
Because he saw the men of the world ploughing their fields, sowing the
Seed, trafficking, huckstering, buying and selling and at the end winning
Nothing but bitterness
For this he was moved to pity.

From the Upāsaka Śila Sūtra.

Translated from the Chinese by Arthur Waley

I suspect (though on the basis of no evidence) that she chose this not so much in relation to the disorder to be described but as reflecting the time (1947) when she was writing, as well as her own character. She had steered the *Treasury of Human Inheritance* through the destruction of two world wars, only to reach a new era of potential world conflict. It must have required a particular combination of serenity, optimism and determination to continue her work undeterred by the surrounding catastrophes.

Conclusion

The *Treasury of Human Inheritance* was (and remains) a unique undertaking, whose importance as foundation for the emerging fields of human and medical genetics has been considerably underestimated. The venture was fortunate to find in Julia Bell a person with the necessary combined clinical and mathematical skills, as well as the ability to write clearly; her long standing involvement (over half a century) with the project gives it, or at least those parts for which she was responsible, a unity that would otherwise have been impossible.

This article has tried to illustrate the numerous aspects of the work that have proved not only original but also of real value to modern genetic studies of inherited disorders. There can be no doubt that Julia Bell was not only a key element in the remarkable development of human genetics at and around University College London in the mid-20th century, but that she was also one of the true founders of the field that we now recognise as medical genetics.

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