Letters to the editor

‘From genetous ailments to genetic disorders: Ireland’s On Idiocy and Imbecility revisited’

SIR—William Ireland’s treatise On Idiocy and Imbecility was one of the first modern textbooks on intellectual disability. It suggested a complex nosography which included a genetous category for currently undefined cases in order to concentrate on the unresolved problems that they represent. The subsequent development of medical genetics has brought a host of answers to Ireland’s questioning. Yet the term genetous could be revived, and contrasted to the term genetic, when there is a need to imply a more partial role of the genome in determining characteristics.

It is 127 years since William Wotherspoon Ireland published his seminal treatise. This influential volume is arguably the first well-organized medical textbook on intellectual disability. While recognizing nosological difficulties, the author suggested a classification including 12 types of intellectual disability which range from ‘inflammatory’ to ‘cretinoid idiocy’. Notably, Ireland defined ‘mongoloid idiocy’ as a particular type, further characterizing the condition which had previously been described by Esquirol, Séguin, Duncan and, notoriously, John Langdon Down, while also endorsing the ‘ethnic’ reference of the latter two.

Terminology has constantly remained a difficult issue in the field of intellectual disability. Some fifty years ago, the opening chapter of another landmark monograph was entitled The Inadequacy of Present-Day Concepts of Mental Deficiency and Mental Illness. Current nosography still shows wide variation between countries, for example, whereas the The International Statistical Classification of Diseases and Related Health Problems revision term ‘mental retardation’ is used with high consistency in North America, ‘learning disability in three domains: environmental, lesion-al, and genetic’. The genetic/genetous distinction might help further qualify the latter.

Among the many innovations presented by Ireland in his book was the remarkable premonition of a ‘genetous’ category defined as follows: ‘At present if we cannot classify some of our cases in a more precise way, we at least may save some confusion by putting them aside from the other classes, and inviting attention to the unresolved problems which they represent’. Although Ireland was a contemporary of Gregor Mendel, and despite his extensive erudition and acquaintance with the French, German, Italian, Spanish, Norse, and Hindustani languages, he was certainly not aware of the abbot’s work on inheritance, which was only rediscovered at the turn of the century. Yet, the term ‘genetous’ appears to be related to the term ‘genetic’ according to the rule for naming chemical compounds where an element has more than one valence. In this system, the lower valence is prefixed ‘-ous’, as in ferrous oxide (FeO), and the higher valence is prefixed ‘-ic’, as in ferric oxide (Fe₂O₃).

The term genetous would appear useful in current-day practice to describe the vast set of conditions where the genome plays a significant but partial role. Most complex disorders (e.g. insulin-dependent diabetes) qualify for this denomination. Linkage studies in affected siblings and genetic association studies are being conducted in a number of conditions to identify the genes that influence disease.

This may open the way to novel preventive and therapeutic strategies. The characterization of the coagulation Factor V Leiden variant in venous thromboembolic disease and of NOD2 polymorphism in Crohn’s disease are emblematic successes of such approaches. It must be stressed, however, that genome liability in these disorders is only partial, an important point that clinicians might intuitively tend to underestimate once mutations are identified.

Monogenic diseases, too, would often be genetous rather than genetic, except for purely Mendelian, fully penetrant disorders (e.g. Tay-Sachs disease). Hereditary breast/ovarian cancer illustrates this point, as only 30–40% of women who carry an inherited mutation in susceptibility gene BRCA1 will not eventually develop cancer. Furthermore, even within the phenotypic spectrum of many monogenic disorders, the terms genetic and genetous might be used to make a distinction between those features that are almost entirely (genetic) or only partially (genetous) determined by the genome. In this sense, in classic cystic fibrosis, for example, pancreatic insufficiency is genetic and pulmonary disease is genetous, as indicated by concordance rates in twins and siblings. This distinction is not redundant to the concepts of penetrance and expressivity, as these do not distinguish between genetic and environmental modifiers. Current clinical and molecular research on autistic syndromes and other conditions where strong genetic and environmental influences are recognized might benefit from this approach. The notion of comorbidity has recently been associated with learning disability in three domains: environmental, lesion-al, and genetic. The genetic/genetous distinction might help further qualify the latter.

Various concepts of comorbidity seemed to guide Ireland in his classification. Accordingly, he provides the reader with carefully detailed recommendations for the management of patients with intellectual disability. Although topical emphases in some of these seem outdated, most reflect refreshing awareness of the importance of integrated treatment with functional objectives, which has now gained general acceptance, and hopefully renders the following observation obsolete: ‘I have seen several cases of club-feet connected with paralysis of certain muscles or sets of muscles, sometimes accompanied by shortening of the bones, in which tendons have been cut by surgeons, especially that class who call themselves pure surgeons because they are entirely free from any knowledge of medicine’. More generally, Ireland’s therapeutic programme prefigures that which was formalized almost a century later by self-named ‘epigenetic’ psychologists, and his approach might be consistent with more recent therapeutic hopes of genetous modulation conveyed by the advances of epigenetics in neurodevelopmental disorders.

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Bernard Dan MD PhD
Marc J Abramowicz MD PhD

Hôpital Universitaire des Enfants Reine Fabiola, Université Libre de Bruxelles
Service de Génétique Médicale, Hôpital Erasme – Université Libre de Bruxelles
‘Quality of life of primary caregivers of children with cerebral palsy: a controlled study with Short Form-36 questionnaire’

We’ve learned a lot. Sometimes I feel like a doctor, therapist, nurse, chauffeur, inventor, builder, computer expert, communication expert... so many things; I sometimes forget I am still a mom.1

SIR–This remark from a mother of a child with cerebral palsy (CP), gives us many clues about the aspects of distress regarding the physical, behavioural, psychological, and social functioning which a parent or caregiver of a child with this disorder is likely to cope with.2

This preliminary study was designed to investigate quality of life status of primary caregivers of children with CP in comparison with primary caregivers of children without CP. In this study, we aimed to investigate two main hypotheses: (1) in contrast to caregivers of children without disability, caregivers of children with CP experience many problems as a result of their higher level of care which they provide, and this will result in lower quality of life scores than expected for caregivers’ age and sex, and (2) some factors will result in a negative impact on quality of life of primary caregivers of children with CP such as, lower motor functional level of the child, job status, and the familial relationship of the primary caregiver with the child.

Participants in the study were 40 primary caregivers (mean age 35 years 10 months, SD 10 years, range 25 years to 60 years) of children with a previous diagnosis of CP who had been followed-up in a specialist CP centre (Spastic Children’s Centre, Faculty of Medicine, Trakya University Hospital, Edirne, Turkey), and were living in the community with their family, and a comparison group of 40 age-matched primary caregivers of children without CP (mean age 34 years 9 months, SD 7 years 4 months, range 19 years to 49 years). Inclusion criteria were that primary caregivers: lived in the community and acted as primary caregiver of a child with CP; did not have any serious or chronic medical disorder (ascertained from history taking) before and/or after the diagnosis of CP in the child; and had not sought medical help for any reason during the last three months. Inclusion criteria for the comparison group were: had no serious or chronic disorder that could interfere with general quality of life; had not sought medical help for any reason in the last three months; acted as the primary caregiver of one or more healthy children without disability. Participants all signed written consent forms, and the study was approved by the University of Trakya Faculty of Medicine ethics committee.

Demographic characteristics of participants such as age, sex, and their relationship to the children (mother, father, or other primary caregiver) were recorded. The relationship between some factors such as primary caregiver’s age and job, and his or her relationship to the child, and Short Form-36 (SF-36) scores were investigated. Children with CP were evaluated using the Gross Motor Function Classification System (GMFCS),2 which is widely used to evaluate abilities and/or limitations arising due to this disorder.

Participants in both groups completed the self-administered SF-36 questionnaires. They were invited to an empty room and were alone while answering questions in order to prevent any bias that might result from being in the presence of an interviewer. The SF-36 health survey has 36 items and measures eight concepts: limitations in physical activity due to health problems; limitations in social activities due to physical or emotional problems; limitations in role activities due to physical health problems; bodily pain; general mental health; limitations in usual role activity due to emotional problems; vitality; and general health perceptions. Higher scores indicate better functioning and well-being.3,4 The Turkish version of SF-36 was translated by Professor Güler Fisek (Bogazici University, Istanbul, Turkey) which was approved by MOS-Trust (originator of SF-36). This approved version has been tested in a study conducted in Turkey and found valid and reliable by Demirsoy.5

Descriptive statistics were used for the evaluation of the demographic data for each group. An independent sample t-test was used to compare the SF-36 scores of primary caregivers and the comparison group.

| Table I: Comparison of SF-36 subscale scores for primary caregivers of children with cerebral palsy and comparison group |
| Subscales | Primary caregivers | Controls | p |
| Mean (SD) | Mean (SD) | |
| Physical functioning | 77.13 (31.37) | 88.28 (11.47) | 0.041* |
| Physical role | 40.00 (46.27) | 56.25 (45.35) | 0.140 |
| Bodily pain | 64.88 (25.14) | 59.70 (21.93) | 0.362 |
| Vitality | 44.25 (15.79) | 60.31 (17.96) | <0.001* |
| General health | 47.65 (22.07) | 60.81 (17.41) | 0.007* |
| Mental health | 55.00 (14.11) | 61.38 (15.07) | 0.053 |
| Emotional role | 35.53 (45.92) | 56.25 (47.47) | 0.047* |
| Social functioning | 65.00 (24.05) | 67.19 (21.71) | 0.690 |

* p<0.05.
Distribution of patients with CP according to the GMFCS levels was not symmetrical i.e. there were more children in levels I and V. Therefore, we divided all the patients with CP into two groups: independent (levels I, II, and III) and dependent (levels IV and V). Influence of these two groups on SF-36 scores of primary caregivers, which was evaluated using a Mann–Whitney U test, showed no significance \((p > 0.05)\).

Thirty-six of the 40 primary caregivers of children with CP were the biological mother (others were: father \(n = 2\), grandmother \(n = 2\)) and 35 of the comparison group were the biological mothers of the child whom they cared for (others were: father \(n = 2\), grandmother \(n = 3\)). There was no significant difference between the two groups regarding age of primary caregivers, age of child, job status, and being the biological mother \((p > 0.05)\).

The SF-36 subscale scores in physical functioning, vitality, general health, and emotional role dimensions were significantly lower in primary caregivers of children with CP when contrasted with the comparison group (Table I).

The results confirm our first hypothesis: primary caregivers of children with CP scored significantly lower than the comparison group in four subscales of the SF-36 health survey questionnaire (physical functioning, vitality, general health, and emotional role). However, the data seem far from confirming the second hypothesis, as we could not demonstrate any significant relationship between the following and the primary caregivers' quality of life scores: lower motor functional level of the child, job status, and familial relationship of the primary caregiver with the child.

Most research in this field has been focused primarily on clinical and/or functional outcomes and possible treatment strategies for patients with CP. We tried to focus our investigation on the primary caregiver, who is one of the most important members of the medical team treating the child with CP.

It is a well known fact that quality of life is subjective, and it should be considered that it may show important differences between countries, cultures etc. However, we believe that having a child with CP may well be deemed a universal problem which has profound shared characteristics experienced by all mothers who are dealing with this problem while living in the community with their child.

Magill-Evans et al. suggest that many previous studies of families which include a member with a disability lacked a comparison group of families without a member with a disability. In this preliminary study, we used a well known, valid, and reliable generic quality of life instrument which is suitable for comparison of general populations. Despite the presence of some limitations which will be discussed below, this controlled design should be considered in the interpretation of the results, especially regarding cross-cultural variations. Overall, quality of life of primary caregivers (biological mother \(n = 36\)) of children with CP was found to be negatively affected, but the level of motor function and/or severity of the disease which was evaluated by GMFCS had no further impact on health of primary caregivers. Once a catastrophic event occurs, its severity seems to have minor importance.

Two limitations of this study should be noted: firstly, types of CP were not taken into account; secondly, some factors closely related to the outcome of the disease process such as presence and/or degree of spasticity, contractures, and other complications were not evaluated. However, a widely used functional scale which may be considered specific to CP was used to determine degree of disability.

In conclusion, the main finding of this study has been to highlight the vital importance of evaluating and, when necessary, treating people who have potential physical, psychological, or social problems related to caregiving that may interfere with their general quality of life. In order to manage the disability of a child with CP effectively, the possible disability of the primary caregivers (and/or the mother) should be considered. We feel that this approach will improve our understanding of many aspects of the rehabilitation of children with CP.

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Hakan Tuna MD Assistant Professor
Halil Ünalan MD Associate Professor
Filiz Tuna MD Assistant Doctor
Siren Kokino MD Professor

*Trakya University, Faculty of Medicine, Physical Medicine and Rehabilitation Department, Edirne, Turkey

Istanbul University, Cerrahpasa Faculty of Medicine, Physical Medicine and Rehabilitation Department, Istanbul, Turkey

Correspondence to:
Dr Hakan Tuna
Assistant Professor
Trakya Universitesi
Tip Fakultesi Fiziksel Tip ve Rehabetasyon AD
22030–Edirne, Turkey
E-Mail: bakantuna@trakya.edu.tr

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