DEVELOPMENT/PHENOTYPE

# The developmental approach to the study of Down syndrome: Contemporary issues in historical perspective

**Tara Flanagan, Natalie Russo, Heidi Flores and Jacob A Burack**

The developmental approach provides an essential framework for understanding Down syndrome. Paradoxically, this framework both narrows and broadens the scope of research in the field. The narrowing involves a more fine-tuned approach to diagnosis, a more precise delineation of skill in relation to specific aetiology and developmental level, and fine-tuned matching strategies that involve comparisons on specific aspects of functioning. The broadening of the scope involves the consideration of the "whole child" in terms of personality, social, and emotional development, within the context of families, communities, and societies. This far-reaching developmental perspective revolutionised the study of intellectual disabilities with its theoretical, methodological, and interpretive innovations, while this more precise approach to the study of persons with intellectual disabilities in turn served to transform developmental theory by challenging, extending, and reconceptualising well-established developmental principles.

The current study of Down syndrome is inherently linked to the advent of the application of the developmental approach to the study of the broader population of intellectual disabilities. The conceptual and methodological innovations co-opted from developmental research and applied to the study of intellectual disabilities were, paradoxically, the catalyst for both a narrowing and a broadening of the scope of research. The narrowing of the scope involved more definitive diagnostic criteria of different aetiologies associated with intellectual disabilities, aetiology-specific conceptualisations of intellectual disabilities, and the precise delineation of skill in relation to developmental level and methods for matching by developmental level (for reviews, see REFS 1-6). The broadening of the scope involved studies in the field of intellectual disabilities research designed to better encompass what Zigler referred to as the "whole child"[7,8]. This included accounting for personality, social, and emotional factors of the child, and for the familial and larger contextual factors, all within the context of syndrome-specific developmentally based research. Paradoxically, both the narrowing and the broadening of the research focus within the field of intellectual disabilities were conceived as a response to traditional views in which persons with intellectual disabilities were considered to be a homogenous group with one or more common defects regardless of aetiology or age (for discussions, see REFS 9,10). Conceptually, at least, discussions of strengths and weaknesses among persons with specific aetiologies at specific ages in specific contexts now supersede monolithic pronouncements of specific deficits. Thus, the outcome of the adoption of a developmental framework is a level of precision of understanding of persons with intellectual disabilities that could not even be anticipated in traditional conceptualisations.

The increased theoretical sophistication, broadened scope, and methodological precision provided by the developmental approach serve to highlight the vastness of the endeavour and the difficulties that are inherent to understanding persons with intellectual disabilities. Within this context, persons with intellectual disabilities must always be considered within the context of their aetiology and developmental level, and in many cases with regard to social, behavioural, emotional, familial, and/or environmental factors. And, the confluence of all these factors must be further considered with regard to the intricacies of the ever changing real world. Thus, the increased specificity of knowledge highlights the futility of attempts to generalise findings across the heterogeneous group that we refer to as persons with intellectual disabilities and clearly the notion of a single grouping or field of research under the title of "intellectual disabilities" seems meaningless.

Yet, this is not a call of despair. Rather, the goal is to promote the vision of research of increased precision that has its origins in 19th century writings by pioneers of aetiological-specific research, such as Langdon Down and William Wetherspoon Ireland, and in 20th century writings by developmental theorists, including Heinz Werner, Edward Zigler, Dante Cicchetti, and their colleagues who promoted the interface of developmental psychology and the study of intellectual disabilities. This approach finds fruition in the 21st century sophistication of experimental technology and empirical methodology in the study of genetics, brain functioning, behaviour, social and interpersonal functioning, and emotional well-being, as well as in the study of the relations among them. The emergence of these increasingly precise approaches to research resonates particularly strongly with the advocates of the

more fine-tuned developmental approach with its emphasis on aetiology- and age-specific research within the context of individuals, their families, and their environments. Through this synergy, the key to understanding the heterogeneous group of persons who fall under the diagnostic heading of intellectual disabilities is a bottom up process with small but fine-tuned and precise empirical "stories" rather than a top-down process with bigger and more general but flawed accounts. In this article, we briefly review essential theoretical, methodological, and interpretative contributions of the developmental approach and attempt to forge a framework for research in the field of intellectual disabilities, as well as of Down syndrome, and to reflect upon how the field of intellectual disabilities research has contributed and transformed the understanding of general developmental processes.

## The theoretical timeline: The developmental framework and intellectual disabilities

The study of cognitive and neuro-cognitive functions or abilities may best highlight the contributions of the developmental approach to intellectual disabilities. These types of functions were the singular focus in the first few decades of scientific research on intellectual disabilities, when most empirical work in the field was characterised by a race to identify the deficit that was the primary cause or marker of reduced intellectual functioning. Those who undertook this frantic search emphasised broad constructs of cognition that were considered to be essential across all domains of cognitive functioning, including cognitive rigidity, memory processes, discrimination learning, and attention, among many others (for a review, see REF 11). With the use of sophisticated experimental paradigms, researchers presented compelling evidence of deficient performance in virtually all of these areas of functioning. Each specific defect was touted as the central cause of intellectual disabilities. Unfortunately, the studies were "fatally" flawed as researchers failed to consider essential and obvious conceptual and methodological issues such as the multiplicity of aetiologies associated with intellectual disabilities, the uniqueness of each with regard to phenotypic expression, the inherent differences in developmental level of functioning between persons with and without intellectual disabilities of the same chronological age, and the non-cognitive factors related to life experiences that are associated with cognitive performance. In critiquing and debunking the various claims of the defect theorists, Zigler and colleagues (e.g., REFS 6,7,8,12) introduced a conceptual approach to intellectual disabilities that would be based on classical developmental theory and would revolutionise the way people with intellectual disabilities were viewed and studied.

## The holistic but precise developmental framework

Consistent with early researchers of intellectual disabilities from the 19th (e.g., REFS 13,14) and 20th centuries (e.g., REFS 15-18), Zigler proposed the 'two group approach to intellectual disabilities' in which persons for whom the cause of intellectual disability was familial should be conceptually differentiated from those for whom intellectual disability could be classified as organic[7]. He argued that intellectual ability is characterised by a bi-modal distribution with one mode falling as expected at an IQ of 100, and a second mode, at the tail end of the impaired side of the normal distribution with a mode of 50. Whereas persons with intellectual disabilities with a familial source were considered to represent the lower end of the normal distribution of intellectual functioning, persons whose intellectual disability had an organic basis, represented the area under the second mode. Accordingly, Zigler proposed that these two groups were qualitatively and quantitatively different from one another. Citing epidemiological work by Dingman and Tarjan[15], he elaborated that persons with familial, but not necessarily those with organic, causes of intellectual disability should follow a similar developmental trajectory as typically developing persons, albeit at a slower rate and to a lower asymptote.

The developmental story for persons with intellectual disabilities associated with organic aetiologies was less clear. Consistent with the universal approaches of traditional developmental theory, they were found to consistently traverse the sequences of development for Piagetian and other cognitive tasks in the same order as was evident among persons with intellectual disabilities with a familial source and among typically developing children (for a review, see REF 19). However, the organic insults were expected to be associated with structures of horizontal development, the relationship across areas of functioning, that were different form those of typically developing persons, so that general indicators of mental age would not necessarily be associated with level of functioning in specific domains[20]. Yet, even here the notion of meaningful development was not abandoned as developmental theorists advocated for meaningful relations across domains of functioning, even when the levels of functioning seemed discordant. For example, Cicchetti and Pogge-Hesse argued that specific examples of organic aetiology provide unique opportunities to expand the study of development as they can be viewed as testing the limits of developmental organisation[21]. Furthermore, the slower pace of development and the discrepancies across domains of functioning that are evident among persons with Down syndrome or other specific aetiologies were cited as unique opportunities to examine the synchronies and asynchronies in development and the extent to which the convergence of aspects of functioning in typically developing persons are real or happenstance[22,23]. Accordingly, the notion of local homologies[24], the relationships over time across tasks that require common underlying capacities, was borrowed from the study of development and applied to the study of persons with intellectual disabilities associated with organic aetiologies[5,25,26]. Within this framework, even apparently discordant performance across domains can be considered within the context of an organised developmental system.

## Methodological issues

### The issue of developmental level

One reason that so many areas of defect were identified over the years was that the target groups of persons with intellectual disabilities were virtually always compared with typically developing persons matched on chronological age (see REF 9). By definition, then, the persons with intellectual disabilities were functioning

at a lower level than those without intellectual disabilities and would be expected to perform worse on any task that was age-appropriate and sufficiently sensitive to differentiate between groups with considerably different levels of functioning. Yet, despite the inevitability of the findings of group differences, the defect theorists cited the impaired performance among the persons with intellectual disabilities as evidence of a core deficit.

In highlighting one example of the extent to which advocates of the defect approach misled the field, Iarocci and Burack and Burack et al., demonstrated that the notion of attention as the core, or at least a central, defect, that was perpetuated from the 1960s through the 1990s was based on series of articles in which matching was exclusively based on chronological ages[9,10]. Accordingly, they argued that the findings of attention deficit would be expected on virtually any area of functioning when chronological age matching is used. In other words, the proponents of the attention defect theory had simply found that "lower functioning persons were functioning at lower levels than higher functioning persons". This, of course, is not at all surprising. Rather, based on developmental theory, a deficit among persons with intellectual disabilities would only be important if it was found in comparisons with typically developing persons matched on some relevant indicator of developmental level, ranging from general measures of mental age to more specific ones that mirror the task. A deficit in this scenario is more likely to indicate an actual impairment because the groups are equated on developmental level. Matching on the basis of developmental level was initiated in the 1960s in studies of developmental disabilities, such as intellectual disabilities[7,8] and autism[27]. Although a few defect theorists persist in their use of chronological age matching, by the 1990s, matching on developmental level had become a hallmark of developmental research in the field of intellectual disabilities (e.g., REFS 28,29,30).

As with many of the other areas of scholarship in the developmental approach to intellectual disabilities, the techniques for matching by developmental level continue to be discussed and developed, and are continuously fine-tuned (e.g., REFS 28,30-33). Among others, Loveland and Kelley and Burack et al., highlight the need to utilise matching is linked to the development of abilities that are pertinent to the specific function or task[34,28]. This strategy minimises the chances that differences in performance between the groups might be an artifact of a specific relative strength or weakness displayed by one of the groups in the area of functioning related to the experimental task (for a related discussion, see REF 24). In order to further reduce the risk of confounds arising from a priori group difference, Mervis and colleagues call for rigorous statistical test criteria in the matching of groups[30,31]. With all this increased sophistication in the methodological approaches to matching, the clear conclusion is that the complexity of the developing organism precludes a single perfect approach to comparing across groups of persons. Rather the goal is to simply utilise strategies that minimise the likelihood that the primary findings are associated with obvious confounds.

In the case of yet another paradox, the discussions of matching by developmental level allow for considerably more precise assessments of the implications of the research findings from various experimental paradigms, but also allow for alternative approaches. For example, Burack and colleagues argue for the use of multiple matching measures in order to allow for the identification of the complex profiles of development across areas of functioning[29,35]. However, this approach does not inherently provide insight about developmental changes over time unless it is varied out across different age groups or in a longitudinal paradigm. In an attempt to better depict the dynamic aspects of the developmental processes, both Jarrold and Brock and Cornish, Scerif and Karmiloff-Smith eschew matching strategies in favour of regression models that are used to chart developmental trajectories[36,37]. This approach is reminiscent of the early work on developmental trajectories of IQ among specific aetiological groups, especially persons with Down syndrome (for a review, see REF 38), and is a useful reminder that the study of intellectual disabilities is inextricably tied to developmental theory. Yet, this approach is limited by the extent to which it useful in the complexities of the continually evolving structure in which each domain, sub-domain, and sub-sub-domain of functioning develops at its own rate but also in relation to each of the others. Furthermore, this approach is less useful in the study of adulthood when developmental trajectories are limited or even flat, but the understanding of specific aspects of functioning or patterns of functioning is still informative for intervention. These benefits and limitations of the various developmentally-based strategies highlight the breadth of the developmental approach and that the choice of methodological strategies needs to be guided by the underlying theory and research questions.

## Conclusions: developmental theory and the study of intellectual disabilities

Cicchetti and Pogge-Hesse suggest that development is a dynamic process that is not adequately captured through the simple static delineations of intact and impaired 'modules'[21]. Rather development unfolds over time in a complex interplay of factors that include, but are not limited to, gene-environment relationships, critical periods for the development of specific areas of functioning such as language, the relationship between and within different areas of functioning, and the impact of compounding delays or differences over the course of an organism's growth.

As the influence of the developmental approach in the study of intellectual and other developmental disabilities increased dramatically by the end of the last century (see REF 39) and into the new one[4], the complexities of understanding developmental trajectories of specific aetiological groups was emphasised conceptually[29,35,40,41] and across areas of research. For example, both Burack, Evans, Klaiman and Iarocci, and Cornish, Scerif and Karmiloff-Smith argued that the study of attention among persons with developmental disabilities needs to be considered within the context of specific aetiology, specific aspect of attentional functioning, specific developmental level, and of changes over time[9,37].

The identification of these types of intricacies of the developmental relations across domains of functioning is central to the notion that cognitive and neurocognitive aspects of functioning are intrinsically

interrelated even if not always temporally synchronous[42,43], and, therefore, the study of specific aetiologies provides a effective lens for general notions of development[23]. For example, Zelazo, Burack, Benedetto and Frye argued that their finding that individuals with Down syndrome, who had mental ages of approximately 5 years, tended to focus on a single state rule in theory of mind and rule use tasks was inconsistent with claims that theory of mind is a domain-specific psychological function but is consistent with the notion of developmental organisation[44]. Within this type of framework, the unique developmental patterns evident among groups of persons with intellectual disabilities, in general, and among those with Down syndrome, are informative about general developmental processes and about the integrity of developmental systems in which the organisation is challenged[3].

The study of persons with intellectual disabilities is a clear example of the basic tenet of the discipline of developmental psychopathology that typical and atypical development is essentially related and mutually informative[45,46]. Officially formalised by Zigler, the developmental approach in the study of intellectual disabilities revolutionised research in the area by both narrowing the scope of research to increase its precision and broadening the scope to include notions of the whole person[7,8]. Zigler's influence continues to resonate in current research on intellectual impairments that considers aetiology-specific variables in conjunction with the contextual factors that impact the functioning of a child. The evolutions and revolutions in research concerning individuals with intellectual impairments highlight the theoretical, methodological, and interpretive innovations that were afforded by the introduction of a developmental framework. In turn, this fine-tuned approach to research among individuals with intellectual impairments served a transformative function with which fundamental aspects of developmental theory are challenged, extended, and even reconceptualised.


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**Correspondence to** Jacob A. Burack, Department of Educational and Counselling Psychology, McGill University, 3700 McTavish Street, Montréal, Québec H3A 1Y2 • e-mail: jake.burack@mcgill.ca.

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