

‘DIAGNOSIS’ OR ‘FUNCTIONAL APPROACH’: IS THIS EVEN A SENSIBLE QUESTION?

Exploring the issues with a multidimensional perspective – recognizing that the answer to the question depends on:
‘What is the question? What do we need to know and what are we going to do with the information?’

BACKGROUND:

- In the field of developmental disability, considerable debate and tension remain regarding the best approaches to thinking about how to engage with and serve children with developmental challenges and their families. In many places, service eligibility is based on diagnosis. This approach puts pressure on **families** to **get** a diagnosis, and on **service providers** to **make** a diagnosis. Among the implications of this challenge are what can become a ‘diagnostic odyssey’ for many families, and create moral/ethical dilemmas for service providers to ‘find’ (assign) a diagnosis in order to support the family to access services.

At the same time, people in our field recognize that there may be clinical, political, sociocultural, ethical, moral, and resource implications of this dilemma, and most of us – whatever our personal views – are frequently involved in discussions and debates about this matter. The concepts presented in the WHO’s ICF framework for health bring a new and more detailed group of interrelated concepts to this discussion, and may help clarify some of the confusion.

- This collaboratively created document aims to set out the ‘issues’ – fairly and comprehensively. Our goal is to offer a global, multidisciplinary worldview. It is hoped that this analysis will provide a clearer basis for discussions and ways to move the field forward.
- There is a logical historical explanation for this dilemma. In most areas of acute care medicine, ‘diagnosis’ is important/essential, because it may lead directly to condition-specific interventions. One example among many is the sudden onset of chest pain: Is it a cardiac issue, an upper GI problem, an acute anxiety attack, an intercostal muscle inflammation...? There is only a small number of very likely ‘diagnoses’ – and making the right one, and instituting the correct management, may literally be life-saving.
- Unlike acute-onset situations, where a small number of ‘causes’ are highly probably, in developmental impairments the range of underlying biological impairments creating a similar phenotype can be very wide. In our field, diagnosis is often imprecise and may take a while to ‘develop’ (i.e., be fully uncovered) based on a child’s development and the appearance of manifestations of a condition. We have a host of diagnostic ‘labels’ with capital letters – CP, ASD, DCD, ID, ODD, ADHD – that seem to be ‘real’ entities. These terms are really descriptions of FUNCTIONAL impairments, described with words that, when capitalized, sound like ‘diseases’ People have referred to this situation as ‘adjectives parading as nouns’! Of course, someone has CP or ASD ‘associated with’ (perhaps even ‘caused by’) some underlying impairment in ‘body structure and function’... but for many of us what is important for intervention, management and counselling of families are the functional impacts of the condition or the ‘disorder’. Of course a precise diagnosis may provide information regarding expecting prognosis and possible course of the condition (whether it is progressive or a static lesion).
- THE PURPOSE OF THIS EXERCISE is to try to:(i) identify the many dimensions of the issues regarding both the ‘diagnosis’ and the ‘functional’ approaches to services, and (ii) enable people to see that these issues are almost always complementary to one another (orthogonal) and not in competition.
- People are encouraged to flesh out the table on the next pages, to add in ideas that bring these issues to life. Our collective perspectives can help to create an approach to the issues, and we can then seek the review and comments of colleagues who have not been involved in the development of the work to this point.

TABLE 1: Outlining issues to be included in a ‘Balanced Scorecard’ of the issues

DIMENSIONS TO CONSIDER	VALUE OF <i>DIAGNOSIS</i>	LIMITATIONS OF <i>DIAGNOSIS</i>	VALUE OF <i>THINKING FUNCTIONALLY</i>	LIMITATIONS OF <i>THINKING FUNCTIONALLY</i>
<p>IMPLICATIONS FOR FAMILIES</p> <ul style="list-style-type: none"> ● ‘explanations’/ clarity ● genetics ● prognosis ● explaining to others ● what else? ● “In many communities, the social expectations are about knowing what is wrong and what the diagnosis is. There can be significant pressures from the wider family, friends and community to pursue this agenda as the biomedical focus is very strong. I have seen this at close quarters in India.” 	<ul style="list-style-type: none"> ● Concrete and tangible, and meaningful in the often-prevailing medical model ● Patient/families feel that the quest of knowing what is “wrong” has ended and now they can focus on treatment. ● Patient/families feels they know what to expect after a diagnosis (e.g., prognosis). ● If the condition is hereditary, genetic counsel can be offered. ● The expected course of condition can help prepare the family for what may lie ahead. ● Addition to support groups ● Receive Social benefits to the family and employment benefits for parents(e.g., for leaving early from work/days off, etc.) ● As a means to identify with a group of individuals, to pursue more information, to become more educated as to what to expect and resources that identify with diagnoses (health care specialists, schools, home care, support groups, sporting events, camps, 	<ul style="list-style-type: none"> ● Diagnosis alone does not convey prognosis about specific aspects of people’s functioning ● Generally leads people to take a ‘deficit-based’ approach (what the child can’t do and their problems) ● Diagnosis may lead people to stop thinking and assessing, and attribute all limitations and issues to the ‘diagnosis’ ● Diagnoses may be associated with blanket prognostications, when there is huge variation in all aspects of people’s lives WITHIN a diagnostic category – hence the need for predictions about the PERSON and not the CONDITION. ● Certain conditions may not fulfill clear-cut clinical and investigative diagnostic criteria and there could be overlap between diagnoses. ● With current “need” for a diagnosis, when a diagnosis cannot be identified families always feel there is something missing. If this was not seen as essential this would give them more peace of mind. ● Diagnosis may generate fear/anxiety based on common 	<ul style="list-style-type: none"> ● Focus on the function and functionality, which encourages a strength-based approach ● Opens the ceiling for possibility of what the outcomes, quality of life, activity/participation could be for their child, and for their family as a whole. ● Allows for a dynamic/evolving approach to intervention based upon the child’s actual function and goals ● From a mental health perspective, invites ‘abilities’ to be considered more widely. E.g., to be witness to all a child/family CAN do ● Functional evaluation leads to early identification of problem and early intervention, not losing time waiting for diagnosis ● Early intervention focuses on functionality, rather than diagnosis; hence referrals can be done on identification of functional disabilities ● May help families understand that the journey 	<ul style="list-style-type: none"> ● We need the right tools to capture functioning in ways that are reliable and valid and meaningful/translatable across cultures. ● Some tools are available: GMFCS, MACS, CFCS, EDACS... but we probably need more (and ones that are valid/stable in the early years of life, when diagnosis may be being pursued). ● Just as ‘diagnosis’ may limit people’s thinking, so too might that happen with a focus just on ‘functioning’ ● Sometimes we can miss treatable condition (comorbidities associated with CP or ASD such as seizures)

	<p>socialization opportunities with other families/children</p> <ul style="list-style-type: none"> • Specific diagnoses are often associated with specific ‘co-morbidities’ (e.g., Down syndrome and myopia or thyroid impairment) • Patient/family understand the underlying pathology that explains their child’s functional impairment • Easier to ‘explain’ to family and community • Access to grants and special education may be limited if a definite diagnosis has not been given. Families may be reliant on the financial support given by the state grants so that their child can access therapy. This is true in our setting in South Africa 	<p>misconceptions, including ‘Dr Google’ e.g. Cerebral Palsy</p> <ul style="list-style-type: none"> • In some communities, diagnosis can lead to social ostracism 	<p>ahead is around living their best life with a condition as opposed to searching for the cure, which often leads to life dissatisfaction and guilt</p> <ul style="list-style-type: none"> • A more ‘positive’ outlook and holistic approach is provided than just a label being used. 	
<p>IMPLICATIONS FOR PROFESSIONALS</p> <ul style="list-style-type: none"> • need for diagnostic services (human, technical) • moral dilemmas (e.g., lying vs hurting families) • prognosis (understand the journey they are planning for, will change priorities) 	<ul style="list-style-type: none"> • Condition-specific pharmacological treatment • Clinical training is often based on a medical model, and so this may make more ‘sense’ to the clinician and the framework they work within (perceived professional ‘safety’). • Condition-based scales/evaluations • Perceived to be more objective, and/ or based on ‘lab tests’. • Power dynamics – only medical professionals can 	<ul style="list-style-type: none"> • Does the imperative for a ‘diagnosis’ put professionals in a moral bind, needing to lie to support families)? • Funding/service policies may directly or indirectly influence rate of diagnosis of a particular condition (as seen with ‘spikes’ in rates of a particular condition following the introduction of new diagnosis-specific initiatives). • Does this lead to an ‘epidemic of diagnosis’ as a result of this pressure? 	<ul style="list-style-type: none"> • ICF model of thinking is a strengths-based approach to health issues, and allows people to ‘rule in’ strengths as well as challenges • Mindset change to consider the abilities of a child/family as equal to, if not more important than, impairments/disabilities. • Therapy interventions target the presenting functional impact of the condition so this is a natural position for therapists to 	<ul style="list-style-type: none"> • People need to know and have access to functional classifications • More are needed – but have to be both reliable and valid • Acceptance by the professionals providing services based on functionality alone due to lack of awareness among professionals requesting the diagnosis. Requires ‘culture change’ within the health system/clinical champions. • Evidence base for many interventions focus on diagnosis rather than function

	<p>make a diagnosis, whereas 'anyone' can look at function.</p> <ul style="list-style-type: none"> • Reimbursement • Ability to work with groups of children and provide interactive opportunities based on diagnosis within an organization (e.g., camps with rehabilitation and appropriate equipment and support available) 	<ul style="list-style-type: none"> • Cost involved in making a precise diagnosis with expensive investigations • 'Lost' time during which interventions can be being offered • I think it is easier to train primary care professionals in identifying functional diagnosis than it is to make them masters in developmental disorders 	<p>think of supporting function and participation</p> <ul style="list-style-type: none"> • Patient/family-centered attention • May promote a new chapter/mindset for reimbursement and blow the lid off of the idea of 'plateauing' • Need champions of this mindset • Prognostication and communication are easier and more accurate with knowledge of functional abilities than with diagnosis alone. 	<ul style="list-style-type: none"> • Need to understand a condition its mechanism and impact through the life course to ensure the interventions are appropriate. For example, supporting mobility in a child with dev delay or Down syndrome may be different than a child with dystonic cerebral palsy. • There is a risk that professionals will stop think laterally, not think of differential diagnosis and miss 'mimic' conditions, especially ones that are 'curable'
<p>IMPLICATIONS FOR SERVICES</p> <ul style="list-style-type: none"> • generic vs. specialized • resource availability • tradition vs. modern (ICF) 	<ul style="list-style-type: none"> • Reimbursement is often based on Dx. • Development of expertise about specific diagnoses • Creation of diagnosis-specific clinical programs leads to creation of expertise in those conditions, including clinical and technical resource development. 	<ul style="list-style-type: none"> • People may feel powerless if there are no diagnosis-specific programs or experts available to them • This may impede parents' search for services. • A major issue in developing world where facilities and resources for a specific diagnosis are limited • Limited number of specialists to make the diagnosis (especially ASD) and the resulting long waiting times to have the diagnosis made before services can be accessed. • 'Re-design' of many diagnostically driven services requires vision, planning, negotiation at various levels including political and inter-organisational 	<ul style="list-style-type: none"> • Generic programs focusing on functionality can accommodate 'everybody' • Access can be widened when there is no need for a 'diagnostic ticket' for entry to the services. • 	<ul style="list-style-type: none"> • Services need to be geared for functionality rather than diagnoses and the provision of infrastructure and manpower • Need service reorganisation to adapt to this way of thinking. Post may be funded for a specific condition, etc., so there may be a challenge around being able to change thinking at the same time as structures and process • Allocation of scarce resources may become difficult: if we just go by functionality, the number of eligible person will become much larger than if we go by diagnosis alone • I see this as service level, especially for the therapists.

		<ul style="list-style-type: none"> ● It may be challenging to attract/retain a workforce with skills appropriate to the caseload ● Experientially, working for a diagnosis specific organisation vs a function-based service may lead people to feel 'out of their depth' in the function focused service (for example, one can have 2 children CFCS IV but the way of working with them is completely different – i.e., one child had dysarthria and another has ASD)... 		
<p>IMPLICATIONS FOR COMMUNITY</p> <ul style="list-style-type: none"> ● epidemiological issues ● planning service needs ● thinking/planning ahead ● funding ● how other systems work (e.g., education, welfare...) 	<ul style="list-style-type: none"> ● Able to explain to others what it is, as society still predominantly understands diagnosis. ● Easier to monitor changes in prevalence of a condition based on preventative strategies that have been implemented, changes to awareness etc. ● May be more straightforward for establishment and interpretation of national registers ● Easier to fundraise in the community for 'cerebral palsy' rather than 'limitations to mobility' – ● Do people find it easier to understand a label? 	<ul style="list-style-type: none"> ● Possible expectation that only with diagnosis can Rx be offered. ● Expectation of diagnosis-specific Rx, when in fact almost none of our Rx is specific to a 'diagnosis' ● Is there something that should be here around stigma etc., that links with diagnosis? Is it more inclusive if we are just considering function that may change with age, environment etc. for all of us, that makes for a more open inclusive approach? 	<ul style="list-style-type: none"> ● Community services inclusively planned for transportation, education, labor conditions ● Community can develop programs for young people – educational, recreational, social support, etc. – based on functional needs (e.g., related to mobility challenges, sensory impairments, etc.) ● Also for planning of transport, housing etc. (e.g., planning housing for 10 adults with cp would not tell you what was needed. Housing for 10 powered mobility users MACS level...etc. would be more informative 	<ul style="list-style-type: none"> ● <i>No one can think of any!</i>

<p>IMPLICATIONS FOR RESEARCH</p> <ul style="list-style-type: none"> ● condition-specific vs. non-categorical ● challenges for funding ● challenges for publication 	<ul style="list-style-type: none"> ● Homogenous groups Funding and how to categorize the call for research in a group of individuals ● Potential ‘sense’ of homogeneity 	<ul style="list-style-type: none"> ● We often cannot find ‘enough’ of the ‘right’ diagnosis... ● Using diagnosis may ‘lull’ us into a false sense of homogeneity when in fact this is not the case. ● Even when the numbers are big enough, we usually stratify by... functioning! (e.g., GMFCS levels for CP) This has led to difficulties in the past, for example, there is good evidence for CIMT, but families and researchers then become confused and think this applies to all children with cp rather than a subgroup. 	<ul style="list-style-type: none"> ● Comparison of associated conditions based on functioning, despite varied diagnosis ● Includes heterogeneity and variability of how children with impairments and disabilities grow and develop (public alert! that really does still happen) 	<ul style="list-style-type: none"> ● Many funders (and reviewers) are still focusing on ‘diagnosis’ – hence the need to explain ‘non-categorical’ concepts – related to function! ● May be a different causal mechanism underlying the same functional presentation, which can result in differences in the nature and magnitude of response to an intervention at the individual level (diluting overall effects)
<p>EXAMPLES OF WHERE THIS ‘WORKS’ OR IS CHALLENGING!</p>	<ul style="list-style-type: none"> ● The ICF blends ‘health condition’ (i.e., diagnosis) with functioning (‘activity’) 		<ul style="list-style-type: none"> ● The increasing availability and use of the functional classification systems speaks to people’s valuing these... does not mean Dx is not also valued, but there is an appreciation of the importance of thinking ‘functionally’ ● National Disability Insurance Scheme in Australia is based upon function not diagnosis ● Therapists are already treating the presentation of a condition. The step to non-categorical thinking is I think therefore very doable. ● Preschool communication intervention programs like Hanen are based on supporting parents’ skill development irrespective of the clinical diagnosis 	<ul style="list-style-type: none"> ● It terms of limitations (in Australia), it does rely on the families having to strongly advocate for their children and focus on their limitations when trying to advocate for funding. The families who focus on the strengths on their children might not get as much funding bc the planners hear they are doing well and therefore think we can cut funds! ● Yes, we (in the UK) can have that problem too. You will only get benefits by highlighting what can’t be done and what is difficult ● This is also true in China Wei

