Course: Introduction and Assessment of Mentally Retarded Children-II

SEMESTER: AUTUMN, 2018

(3604)

ASSIGNMENT# 2

Q. 1 How the assessment of vocational aptitude and competency car be carried out by the teachers of children with intellectual disability?

ANS: The National Education Program is further elaborated in the Strategic Plan of the Ministry of National Education with the formulation of the National Education Vision, namely Building Intelligent and Competitive Indonesian People. To achieve this vision and mission, the Ministry of Education launched the Three Pillars of general policy on national education development, namely as follows: (1) Increasing equity and expanding access to education, (2) Improving quality, relevance and competitiveness of education, and (3) Strengthening management, accountability, and public image of education management. The implication is that policies and program development for all elements in the national education system must describe the three pillars according to the type and level of education. Referring to article 15 of the Indonesian Education System Law No. 20 of 2003, this type of education includes general, vocational, academic, professional, vocational, religious and special education.

The type of education that is the orientation of the discussion in this study is specific to Vocational Education. Puyate says that for the vocational programs to be fully implemented, students as stakeholders have to be made aware of such programs and their importance; they must become interested in practical skills-oriented lectures, and cognitive skills at the same time .Vocational Education is secondary education in the form of Vocational High Schools (Sekolah Menengah Kejuruan/SMK) or Vocational Madrasah Aliyah (Madrasah Aliyah Kejuruan/MAK) which prepares students especially to work in certain fields. are schools in secondary education that specifically prepare their students to be skilled, ready to work, adaptable to the environment and change, and able to develop themselves according to the needs of the workforce.

SMK as one of the education levels is expected to prepare graduates quality too. For this reason, in their education, vocational students are equipped with knowledge, attitude, skills and life skills that are useful for himself and the community. In order to provide life skills, vocational schools work with the business world / industrial world as a partner institution.

After attending education at a vocational school, armed with knowledge, attitude, and skills in their fields, SMK graduates are expected to be able to choose various channels life. Realizing this, the role of vocational schools is truly education a terminal that connects various dimensions of interest, both government, community, and graduates themselves.

2. Teorical Framework

2.1. Vocational Education

The term vocational education is general and includes every form of education that aims to the acquirement of qualifications related to a certain profession, art or employment or that provides the necessary training and the appropriate skills as well as technical knowledge, so that students are able to exercise a profession, art or activity, independently of their age and their training level, even if the training program contains also elements of general education ·Principles of vocational education are defined as generalizations that state a preferred practice and serve as guidelines for program and curriculum construction, evaluation, selection of instructional practices, and policy development ·

A course of study that counts as vocational education in one country may be a part of higher education in another, and it may be entirely absent in a third country because the proffesional domains and hierarchies are

organised differently, but also takes place in different parts of educational system ·Guo & Lamb says that vocational educatiob which refers to the formal vocational education and training provided by vocational schools and training centress established bt trade administrative bodies, trade organizations and their subordinate enterprises ·

Creating high-level specialists is a continuous process, which shouldn't end with a diploma of basic vocational education and demands of an employee to participate in further educational programs on either behalf of his or her employer or at his or her own expense ·

Technical and vocational educations are one of those ways by which a skillfully trained and graduated person becomes ready to commit and enter any related business. To help the efficiency and empowerment of technical and vocational educations has various dimensions among which internal and external function can be pointed out ·

Martins et all says that the vocational education and curriculum differentiation as a strategy to reduce the school early leaving and to the acquisition of the relevant skills needed in this "knowledge society" · Vocational education, has, therefore, been thought of as a "wise business investment" both for nation and the individual. Vocational education in its broadest where sense pertains to all occupations and all people ·

Vocational Education and Training (VET) is organized around a principle of dual education where the apprentices alternate between a vocational school and training place in a company or firm. Thus the provision of a sufficient number of training places is the main precondition for the success of the system [10]. The growth of the number of people with secondary vocational education, the improvement of the level of economic development of the region, and the increase of the share of the employed in the manufacturing sector help reduce the rate of youth unemployment ·Vocational education in a changing world is undergoing a particular process of transition encompassing policies, practices and concepts. Certaninly, such transition processes do not happen whitout human intervention ·The bottom line is that vocational education is responsible for the basic skills in the work place. More improtantly, humas need more education, especially vocational education for work ·

2.2. Assessment

Assessment in education must, first and foremost, serve the purpose of supporting learning. So it is fitting to start a study assessment with an exploration of the meaning and practices od assessment which serve this purpose most directly Assessment exist to promote learning and to inform others about what has been successfully learned William says that the word "assessment" was used primarily to describe processes of evaluating the effectiveness of sequences of instructional activities when the sequence was completed Assessment is vital to the education process. In schools, the most visible assessments are summative. Summative assessments are used to measure what students have learnt at the end of a unit, to promote students, to ensure they have met required standards on the way to earning certification for school completion or to enter certain occupations, or as a method for selecting students for entry into further education ·

Assessment as part of classroom activities is a fundamental process required to promote learning and ultimately achievement ·Consequently, evaluation takes on different characteristics and is interpreted differently in formative versus summative situations. This distinction was soon applied to the assessment of students. Specifically, formative assessment was defined as occurring while knowledge is being learned. Sumative assessment was defined as occurring at the end of a learning episode—for example, at the end of a course ·The diversity of assessment methods utilized is broad. The range includes the preparation of student profiles and portfolios, structured teacher ratings of student capabilities demonstrated in the course of regular classroom work, evaluated student projects, and even organized competitive events ·Furtermore, Oz says assessment approach associated with constructivism, with numerous terms used almost interchangeably to mean similar practices and procedures, including terms such as formative assessment, teacher-based assessment, classroom-based assessment, school-based assessment, dynamic assessment, and alternative assessment [21].

The term performance is used to refer to knowledge or skill aquired through instruction or study as well as the process of acquiring knowledge or skill through instruction or study. The term performance is used to refer to things accomplishment of things (process) · Assessment data are systematically collected concerning the student's interests, aptitudes, special needs, learning styles, work habits and behavior, personal and social skills,

values and attitudes toward work, and work tolerance ·Three recurring emphases in the literature on assessment for enhancing learning are (a) clarity in illuminating standards that may articulate the gap between what was achieved and what can be gained, (b) the importance of assessment design in prompting and sustaining students' desired learning, and (c) giving students feedback that enables them to improve their learning ·The significance and the value of the adoption of alternative forms of assessment, such as student self-evaluation and portfolio assessment have been described. One of the central aims in education is the enhancement of students' abilities as learners. The assessment approaches therefore need to support this aim. Teachers need to be aware of the interrelationship of pedagogy, assessment and the curriculum. They need to change their teaching practice accordingly in the adoption of alternative assessment approaches ·

Any assessment strategy that aims to be inclusive should deploy a variety of methods for assessment (for example written assignments, presentations, reflective accounts and so on), so that the same students are not always disadvantaged. All participants need to be provided with equivalent opportunities to demonstrate their abilities and maximize their potential ·

2.3. Competency

The measurement of student performance is a complex process and will generate throught provoking and controversial disscussion. Other believ that since education is a state fuction, the state should be responsible for measuring, assessing, and certifyng possession of competencies ·Competency as a knowledge, skill or attitude that enables one to effectively perform the activity of a given occupation or function to the standards ecpected in emplyment ·Competency also called dimensions, a group of behaviors that are specific, observable and verifiable, that can be reliably and logically classified together and that are related to job success. Competency areas have both a little and a definition. The little identifies the broad category of the competency and the definition lists knowledge, skills, and abilities of other aspects of character or performance required to be competent ·

The term "competency" has not been clearly defined in the literature. Two main meanings of the term have been identified, one referring to the outputs, or results of training – that is, competent performance ·Competency assessment system is made up of levels with performance indicators and assessor guidance on interpreting the indicators, and a suite of assessment tools including assessment plans, evidence-gathering templates, skills matrices for recording the summary outcome of all assessments, and categorisation flowcharts to help allocate work ·Furthermore, Wright says that competency assessment is a way to articulate the ever changing expectations of the job and the organizational vision ·Competency assessment and the first wave of report to management usually signal the departure of the consultans the conclusion of the project ·Often, individual competencies are developed through experiences, circumstances, environment, and to an extent, chance ·

3. Method of Research

This research using a qualitative method because is one method of research that offers design exploratory research aiming. Unlike the design of experimental research for example on the design of qualitative research investigators did not starts from a certain frame of mind, but let the natural setting of research / as they are and seeks to understand the phenomenon that is by putting yourself in the object being studied (empathy) ·

This study aims to describe the application of performance assessments in the learning process of skin beauty program students in Vocational High Schools, describing the application of performance assessments in the competency test of students of skin beauty programs in vocational schools conducted by the association of beauty experts from the Beauty Professional Certification Institute, and knowing the impact of application performance assessment on the learning process and competency tests on the results of recommendations for professional competency certification for students in vocational schools, which are recognized nationally and cross-sectorally as professional workers from the National Professional Certification Agency. Therefore, as is usual in the step of scientific research conducted by researchers is to explore (exploration) of the object under investigation. Data is collected from an experienced background (natural setting) as a data source directly. The meaning of data can only be done if the depth is obtained from the facts obtained. This approach is determined by observing the phenomena observed by the subject of the conceptual world through actions and thoughts to understand the meanings compiled by the subject around everyday events ·

Field research was conducted on 4 Vocational Schools selected based on purposive sampling with certain criteria in the period from July to December 2012. In this study the location was determined by the following criteria: Vocational School that opened a skin beauty program, at least 2 teachers who had been certified as competency assessors beauty, the practice room meets the criteria as a place for competency testing, vocational schools are able to provide competency certification budget for students, vocational schools include locations in the provincial and city / district capital regions.

This study uses multiple case study methods because all elements in the research focus are related. There are three phases of activity namely research design, single case data collection and analysis, and cross case data analysis.

4. Result of Research

This study begins with a document study to determine the sample, which is first looking for data on how many Vocational Schools open the Beauty Management field in Indonesia, secondly determining how many Vocational Schools in the Beauty Management field meet the criteria in this study. Furthermore, the researchers contacted Vocational Schools that met the criteria in this study to request their availability as research samples. In this study, to determine vocational school as a research location, researchers used purposive sampling method. Determination of research locations based on the following criteria: (1) Having a teacher with the following criteria: the number of teachers in the field of beauty at least 2 teachers in each class, teachers with an undergraduate education background in the field of beauty expertise of at least two people each vocational school, teachers have minimal teaching experience 3 years in the field of beauty, the teacher has a certificate as a competency assessor of at least 2 people in each vocational school, (2) the school facilities that must be available are: theory study rooms, practice rooms, cosmetics / practical materials, and standard practice tools, (3) The infrastructure in question is: schools have easy access to public transport vehicles or private vehicles, schools have adequate electricity facilities, schools have clean water and dirty water sources and channels, and schools have communication facilities (telephone, iphone) and internet networks .

Focus I: Description of the Application of Performance Assessment on Document Data Learning Process at SMK 3 Bogor

The learning process document data reviewed in this study are grouped as document data derived from teachers and documents produced by students in the learning process.

1) Findings of Learning Document Data for Teachers of SMK 3 Bogor

a). Learning Process Draft (RPP)

Teachers of SMK 3 Bogor have prepared learning documents within one semester.

b). Teaching material or Reference.

Teachers of SMK 3 Bogor have used teaching materials or references which include display material in the form of presentation texts and various forms of poster images. While the references used are text books published by the Directorate of Vocational Education, and modules obtained by teachers from the Beauty Teacher training training from PPPPTK Business and Tourism, as well as several textbooks published by national publishers and several references from magazines.

c). Practical tools and materials.

The practical tools that will be needed for learning have been arranged in packages to be used in every learning activity by the teacher of SMK 3 Bogor when demonstrating work procedures. While the practice material has been planned and prepared by the team at the beginning of the semester and its use must be through the application of each practice according to the type of practice and number of students.

d). Job sheet.

Job sheet format that contains procedures for practice activities, and is distributed to students in accordance with the competencies practiced.

2) Findings of Learning Documents for Students of SMK 3 Bogor.

Learning documents prepared by students of SMK 3 Bogor are documents that are useful as evidence of learning, namely in the form of Job sheets, Porto folios, and Project work on the competence to treat facial skin without problems manually.

Job sheet

A collection of job sheet students from SMKN 3 Bogor is a series of practical activities in the classroom under the supervision of the teacher. Student job sheets contain a plan for a series of practical activities ranging from preparation of tools, materials and cosmetics, implementation steps, and post-practice packaging.
In making student job sheets, the teacher has helped shape the performance pattern so that students just add the variants used in relation to the type of equipment used to treat the face and the type of facial care cosmetics, as well as the size and method of application. Thus the job sheet of students of SMKN 3 Bogor is in accordance with the rules of mastery of competence in this study, only the student job sheet is still made in handwriting.
The teacher has commented on the student's job sheet, and if there are performance criteria that have not been implemented then given a sign for further improvement or training.
Portopolio
A portfolio of practical tasks for students of SMK 3 Bogor in the form of a collection of practical work reports at school, at an internship place. In Porto folio students of SMK 3 Bogor also included evidence of student involvement in various practical activities related to the application of competencies in the form of certificates or certificates.
Project work.
Project work of students of SMK 3 Bogor is in the form of a final assignment given by the teacher which is related to the application of the business concept as a business in the skin beauty services industry.
In carrying out this task students of SMK 3 Bogor, in groups of 2 to 3 people, and are targeted to carry out care services at least 5 times. Students are first directed to make business proposals that take into account capital and selling prices or profit targets. Each group worked on the concept of a beauty service business with a duration of two weeks. Reports Project business results are delivered in class by each group. On average the group gets three people, only one group gets customers up to 6 to 7 people. As a reward for the group that gets the most customers, the teacher gives awards in the form of congratulations from the teacher and friends. Project service business capital is from the teacher, it must be returned to the teacher, after being calculated, 30% is a service for students.
3) Findings of Observation Data on Learning Process at SMK 3 Bogor.
Technically, the observation of the learning process is done through the replay of the learning video with the consideration that the teacher in charge of teaching can watch to do self-reflection. Other teachers are asked to observe the learning process that has been carried out by one of their colleagues and prepare to provide input to peer teachers as part of a constructive criticism process.
The description of the findings data from the observation of the sub focus of learning at SMK 3 Bogor was conducted on theoretical learning and practical learning as follows:
Based on video recordings, it appears that the theory learning process is carried out in the SMKN 3 Bogor practice room. The teacher prepares the tools, cosmetics that are arranged according to the order of use and subject matter to be explained. The teacher conditions the class by praying first. Then open with greetings to begin learning. Then the teacher presented the subject matter with the demonstration method. Students sit freely on the floor watching the teacher's explanation. Students are welcome to ask if something is still unclear or not understood.
From the table above it can be described that in the 135 minute time allocation, the average observations of the four observers: the average time allocated for activities by the teacher was 78.75 minutes, the allocation of student activity time averaged 31 minutes and the time allocation for silence in class created an average of 25.25 minutes.
Teacher activities include:
Activity 1, in acknowledging the element of 4.25 minutes, this indicates that the teacher has not been responsive in giving recognition of the students' performance or knowledge in verbal learning.
In activity 2, encouraging, that is to motivate students in learning the allocation of time used 6.25. minute.
Activity 3, accepting, in this case the teacher accepts proposals, opinions or comments from students in the learning process, the teacher allocates 7.25 minutes
Activity 4, asked, was an activity to confirm to students the material presented, the teacher allocated 8.25 minutes.

Activity 5, explained, the teacher needed 19 minutes to deliver the introductory material using the power point impressions.
Activity 6, giving instructions, allocated time 13.75, the teacher gives additional information when students ask.
Activity 7, time allocation of 15 minutes. In this case the teacher demonstrates practice procedures that are considered unclear for students after viewing the video show.
Activity 8, shows authority, the teacher takes 5 minutes. Teacher activities to admonish students who are not cooperative or interfere with the learning process, or students take harmful actions.
While Student Activities include:
Activity 1, reacting to the average individual observer seeing the reaction of students is 14 minutes. This reaction is in the form of asking, giving opinions, choosing and identifying practice tools.
Activity 2, reacting the group requires 9.75 minutes.
Activity 3, for students is an individual initiative allocated 7.25 minutes, shown to some students who actively form the preparatory working group. Silence practice is no physical activity of the teacher and students, this position is quiet classrooms because the teacher and students are watching the process of taking care of the surrounding face 25.25 minutes
The implementation of practical and theoretical learning has a span of one week, therefore students are expected to be able to solidify the understanding of the theoretical material that they have acquired and practice caring skills before practicing at school. Students get the APL 02 format from LSP-BNSP which contains assessment criteria for taking care of faces that must be mastered, and must fill in the check list form as a self assessment process. This process is to implement the principles at the fair performance assessment. The following table is a summary of the results of observations of the activities of teachers and students in learning the practice of competency in treating facial skin without problems manually with an allocation of learning time of 135 minutes.
From Table 2, it can be described that in the 135 minute time allocation, the average results of the four observers' observations were for the allocation of activities carried out by the teacher by 44 minutes (32.6%), and for the allocation of student activity time by 91 minutes (67.4%). This indicates that in the process of learning the practice of competency to take care of facial skin without problems manually in SMK 3 Bogor, the teacher's function is more at facilitating students' practice, student activities appear more active by comparison.

No	Category	*Observer*				Average
		1	2	3	4	
A	Teacher activities					
	1. Open the lesson	3	2	3	3	2.75
	2. Do a check list of student activities in preparation	3	3	2	2	2.5
	3. Conduct a check list of student activities in the implementation of care	20	23	22	23	22
	4. Give instructions	2	3	4	3	3
	5. Demonstrate	2	3	3	2	2.5
	6. Stop the practice if it is dangerous	3	3	2	2	2.5
	7. Provide feedback	8	8	9	7	8
	Sub Total	41	45	45	45	44
B	Student activities					
	Preparing: areas, lenna, tools and materials, personal and safe environment	4	3	3	3	3.25
	Consult and Customer Preparation	4	3	3	4	3.5
	Perform Skin Analysis	5	3	3	4	3.75
	Face Carry out care Face skin	70	70	68	68	69
	Provide advice and advice after treatment	6	5	6	5	5.5
	Clearing work areas, tools, materials and cosmetics	5	6	7	6	6
	Sub Total	94	90	90	90	91.

There are 6 elements of performance from the competency of taking care of the face, not having problems manually, which must be mastered by students in the practical learning process at SMK 3 Bogor, namely the elements of preparation, customer consultation, skin analysis, skin care, advice and advice, and packing up work tools.

5. Discussions

The results of the study show that it is true and proven that the application of the performance assessment system carried out in the Learning Process of competency in treating facial skin is not problematic in vocational students in the Skin Beauty program in four locations, the achievement of competency learning is fulfilled. In the Competency Test to treat facial skin is not a problem for students Skin Beauty program in 4 Vocational research locations conducted by the beauty professional certification body (LSP) beauty, students can pass well and the results can improve the quality of these SMK graduates so that they obtain professional certification from National Professional Certification Agency.

Q. 2 Can we consider Autism Spectrum Disorder as a variant of mental retardation? Provide justifications for your answer.

ANS: Autism spectrum disorders

Autism, also referred to as autistic disorder (designated as OMIM by Mendelian Inheritance in Man, an database of human genes and genetic phenotypcs), is a developmental neuropsychiatrie disorder that was first described in 1943 by Dr Leo Kanner. DSM-IV *(Diagnostic and Statistical. Manual, 4th Edition,* American Psychiatric Association), or similar criteria are used for diagnosis of autism and other disorders in this spectrum, referred to as the pervasive developmental disorders (FDD). DSM-IV criteria for autism include onset by age 3, impairments in social interaction and in social communication, as well as repetitive and stereotypic patterns of

behaviors or restricted interests. Asperger syndrome represents a higher-functioning form of ASD. Pervasive developmental disorder-not otherwise specified (PDD-NOS) also involves deficits in all three domains (social interaction, social communication, characteristic behaviors/interests), but the deficits do not reach threshold criteria for an autism diagnosis. In this review, Asperger syndrome, and PDD-NOS will be referred to collectively as autism spectrum disorders (ASDs).

Epidemiological twin studies of ASDs

ASD twin studies have shown that the concordance rate for monozygotic (MZ) and dizygotic (DZ) twins ascertained for ASD differed substantially, with an MZ:DZ ratio of approximately 10 to 1In these studies, estimates of heritability (ie, the proportion of the variance explained by genetic causes) in ASDs were above 93%, with little or no evidence for shared or nonshared environmental factors. Th ese conclusions agree with multiple, independent family studies, which show a very strong aggregation of ASDs within families, consistent with a genetic etiology for ASDs, with little support for environmental factors (eg, ref 6). Note, however, that risk for ASD is not transmitted in a simple manner. This is exemplified both in the family studies (eg, ref 6) and in the genetic conditions that can result in an ASD diagnosis, which are typically non-Mendelian and include X-linked inheritance, de novo mutations, and chromosomal alterations, as well as transmission of unbalanced translocations .

Common variant and multiple rare variant hypotheses

In common genetic diseases, such as ASDs, there have been two models for genetic etiology, which are not necessarily mutually exclusive. These are the common disease/common variant (CD/CV) and the common disease/rare variant (CD/RV) models (the latter is also called the multiple rare variant/MRV model). CVs are genetic variants that are found to be more widely distributed in the population and are associated with a modest increase in risk, and typically associated with odds ratios (ORs) less than 2 and often with ORs in the 1.1-1.3 range. The interaction of a CV with other CVs and with other factors can act together to increase risk. In contrast, in the MRV model a large number of rare, and even very rare, variants underlie the disorder. These RVs are typically associated with ORs that can be quite substantial, and may contribute the major part of the susceptibility for a given individual. At the extreme, RVs have high ORs and are equivalent to rare deleterious mutations. Population attributable risk (PAR, ie, the proportion of disease in a population that can be attributed to a given etiological agent) can be quite high for CVs, irrespective of their lower ORs, as the variants are more widespread, while RVs, even with high ORs, will have a low PAR when considered individually. With M RVs, PAR becomes significant when considering the RVs together. CVs and RVs will be treated very differently in the clinical setting, and also require alternative approaches for their discovery. For these reasons, understanding which mechanism(s) underlie a given disease entity can have important ramifications. Tile CD/CV hypothesis had been challenged from a population genetic perspective and, as we will note below, there was and is increasing evidence that ASDs can result from RVs, leading to a M'RV model to explain much of the ASD risk.

Methods for identifying genetic loci for ASDs

Genomic variations leading to large-scale deletion and duplications associated with ASDs were first identified by karyotyping (eg, ref 11). More recently, the use of genome-wide arrays to query for copy number variants (CNVs) has identified additional genomic variations associated with ASDs (see below). As these methods evolve and their resolution increases, additional genomic imbalances associated with ASD will certainly be identified. Similarly, the search for both single-base RVs and CVs in disease has also been profoundly affected by the evolution of technology. In the past, such studies focused on CVs or RVs in candidate genes, identified based on biological grounds and/or positional information following linkage analyses (eg, ref 12). With the evolution of genome-wide single-nucleotide polymorphism (SNP) genotyping, the focus on CV SNPs that increase risk for disease has shifted to genomewide association analyses (GWAS). Similarly, with the development of high-throughput sequencing, hundreds of genes can be sequenced for RVs in ASDs. In the following sections, we will review RVs associated with ASDs, including genetic conditions, CNVs, and mutations

Multiple rare variants in autism spectrum disorders (ASDs). While epidemiologically rigorous studies have yet to be carried out, there are reasonable estimates for the prevalence of some of the genetic

contributors to ASDs. Some of the more common ones are shown here, together with some of the rare variants identified in recent studies.

Genetic etiology	Estimated prevalence in ASD
Karyotype abnormalities	5-10%
Fragile X syndrome	2%
15q abnormalities	2%
Tuberous sclerosis	1%
16p11 deletions	1%
22q13 deletions/*SHANK3* abnormalities	0,75%
22q11 abnormalities	0,50%
Rett syndrome	0,10%
c30rf58 deletions, homozygous	Rare
CNTN4 deletions	Rare
DPP10 deletions	Rare
DPP6 Deletions	Rare
NHE9 deletions, homozygous	Rare
NLGN deletions/mutations	Rare
NRXN1 deletions	Rare
PCDH10 deletions, homozygous	Rare
PCDH9 deletions	Rare
PTEN mutations	Rare

Genetic conditions associated with ASDs

A variety of genetic conditions, of which most can be syndromic (ie, associated with recognizable clinical signs, including dysmorphic, metabolic, or neurological features) can present with ASDs. These include 15qll-13 duplications, 22qll deletion/DiGcorgc syndrome, 22qll duplication syndrome, 22ql3 deletion syndrome, adenylosuccinate lyase deficiency, Angelman syndrome (AS), Cohen syndrome, Down syndrome, Fragile X syndrome, MA'CP2-rclatcd disorders, neurofibromatosis, untreated phenylketonuria, Potocki-Lupski syndrome, Prader-Willi syndrome (PWS), FT'/tW-associated syndromes, San Filippo syndrome, Smith-Magenis syndrome, SmithLemli-Opitz syndrome, Sotos syndrome, tuberous sclerosis, and Williams syndrome. For individuals with these syndromes, a proportion of cases can have an ASD diagnosisand for some of these conditions, there have been examples of individuals identified with a primary diagnosis of ASD, and only later was the syndrome identified (that is to say that a proportion of individuals with assessed with idiopathic ASDs may have some of these conditions, perhaps without a typical syndromal presentation).

In some cases, the genetics of the ASD-associated syndrome is well understood. Fragile X syndrome (FXS), caused by a trinucleotide repeat expansion in the fragile X mental retardation 1 (FMRl) gene at Xq27.3, is among the most common syndromes associated with ASDs. Among individuals with FXS, ASD symptoms occur in one quarter to one third of subjects, while the prevalence of FXS is estimated to be 2% among individuals identified with an ASD. AS and PWS, as well as 15qll-13 duplications, collectively are also common ASD-associated syndromes, of which each has different molecular etiology. Rett syndrome, listed among the PDDs in DSM-IV, is caused by mutation in the gene encoding methyl -CpG-binding protein-2 (MECP2) and a proportion of girls identified with an ASD are found to have Rett syndrome.

Novel CNVs associated with ASDs identified by genome-wide scanning

Novel CNVs in ASDs

Whole genome scans for CNVs use genome-wide arraybased methods to search for deletions and duplications. 'ITtiis approach complements karyotyping and targeted methods such as fluorescence in situ hybridization (FISH). There have been several such studies in idiopathic ASDs in the past year, along with additional such studies in syndromal ASDs. Several general findings are worth noting (see ref 33 for a more detailed summary). First, there are increased rates of de novo CNVs in ASDs, particularly in simplex families, reaffirming what was clear from medical conditions associated with ASDs, ie, that de novo changes are significant factors in ASD. Second, there appear to be increases in the numbers of de novo CNVs in the syndromal cases. Third, amongst inherited CNVs, there were individuals (parents or sibs) with the CNV without an apparent diagnosis, consistent with variable expressivity of many known genetic disorders. There were even families where a likely causal CNV was found in one affected child but not in another, suggesting independent etiologies. Finally, there were some CNVs that were recurrent (see below) but there were some CNVs that appeared likely to be etiologically significant but that were identified only once. Algorithms are being developed by molecular cytogeneticists to weight such nonrecurrent CNVs to estimate the likelihood that they are etiologically relevant, considering such factors as size of the CNV, whether it is a deletion or duplication, de novo or inherited origin, gene content, and overlap with known genetic disorders.

CNTN4

Disruption of *CNTN4,* coding for the CAM' contactin 4 which is involved in the formation, maintenance, and plasticity of neuronal networks, has been shown to be a likely cause for cognitive aspects of 3p deletion syndrome, which presents with developmental delay. Recently, deletions in cases with idiopathic ASDs indentified CNVs at the *CNTN4* locus in two unrelated individuals

NRXN1

The first large, genome-wide SNP microarray study (using earlier generation arrays and hence just 10 000 SNPs) was conducted in over 1000 ASDs families by the Autism Genome Project (AGP) Consortium. With stringent filtering, a total of 254 CNVs were identified as being most relevant to ASD. The AGP identified two female sibs with ASD harboring identical de novo deletions at 2pl6, over a portion of the neurexin 1 *(NRXN1)* gene. Additional groups have since confirmed a role for *NRXN1* deletions in ASD. Neurexins function in the vertebrate nervous system as CAMs with critical roles in synaptogenetis and bind to neuroligins, which represent another family of ASD genes (see below).

16p11 CNVs

Another interesting CNV in ASD is in the 16pll region, which occurs in up to 1% of subjects with ASDs. First reported in the studies by Sebat et al and Christian et al, this CNV was also identified in a screen by Weiss et al, in which they investigated 751 multiplex ASD families using 500 000 SNP markers and found a recurrent 16pll deletion (note that some of these studies relied in part on the Autism Genetic Reource Exchange (AGRE) collection and were hence not fully independent) The CNV has recurrent break points defined by low copy repeats and includes - 30 genes, and multiple studies are being carried out in multiple labs to understand whether this represents a contiguous gene syndrome or whether one or two genes in the interval are responsible for the phenotype.

The synaptic genes *DPP6, DPP10,* and *PCDH9*

An additional SNP microarray study using 500 000 SNP markers investigated 427 ASD families.[31] This study described many potentially interesting CNVs (277 CNVs in 44% of ASD families) (including the 16pll deletion). Genes within those CNVs included the synaptic genes *SHANKS, NLGN4,* and *NRXN1* (see above and below) and additional synaptic genes, including *DPP6, DPP10,* and *PCDH9.* The dipeptidyl peptidases (DPP) DPP6 and DPP10, which actually lack DPP activity and have therefore been proposed to be renamed DPP-like, complex with Kv4 potassium channels and potassium-channel interacting proteins (KChlPs) to regulate channel activity. DPP6 and DPP10 are hence important regulators of neuronal excitability, particularly as related to the regulation of firing frequency, integration of signal across dendrites, and neuronal plasticity. *PCDH9* codes for protocadherin 9, a member of the cadherin family of homotypic CAMs, which shows localized expression in particular cortical and thalamic regions in development.

Homozygous deletions in *PCDH10, DAI1* , and *NHE9*

Recently, homozygosity mapping was used to identify a novel large homozygous deletion at 3q24 implicating the *c3orf58* locus (or deleted in autism 1, *DIA1),* which encodes a protein localized to the Golgi apparatus, and a homozygous deletion at 4q28 implicating the protocadherin 10 *(PCDH10)* locus, which encodes a cadherin superfamily protein essential for normal forebrain axon outgrowth. Gene expression studies in rat neurons showed that expression of these genes is regulated by neuronal activity and hence may be involved in synaptic changes related to learning. A gene adjacent to *DIA1,* the Na+/H+ exchanger 9*(NHE9)* encoding a membrane protein that exchanges intracellular H+ for extracellular Na+, was identified with a loss-of -function mutation in autism patients with unrelated parents.

Novel mutations associated with ASDs

SHANK3

The 22ql3 deletion syndrome is characterized by global developmental delay, hypotonia, delayed or absence of speech, normal to accelerated growth and head circumference, mild dysmorphic face, and ASD-like behaviors, and there is good evidence, based on the presence of a recurrent breakpoint, that *SHANKS* is the critical gene in this syndrome. A recent study asked whether mutations in *SHANKS* or chromosomal changes at the *SHANKS* locus were directly associated with idiopathic ASDs, making use of FISH analysis and/or direct sequencing in about 300 cases. The study identified three families with ASD that showed alterations: a family with a proband carrying a de novo deletion in intron 8, removing 142 kb of 22ql3; a family with two affected brothers with a nucleotide insertion, creating a frameshift that modified the C-terminal sequence of the protein (derived from germline mosaicism in the mother); and, a family with both monosomy (resulting in autism) or trisomy of 22ql3 (resulting in Asperger syndrome) in affected siblings, both arising from a paternal translocation. A subsequent study found one de novo *SHANKS* mutation and two 22ql3 deletions in 400 ASD subjects (and an additional deletion in a different cohort). As in the first study, one deletion arose from a paternal translocation, resulting in a child with monosomy at the *SHANKS* locus and a child with trisomy at this locus. 'I lie monosomy was associated with autism, while the monosomy was associated with attention-deficit hyperactivity disorder (ADHD). A recent study found one de novo deletion and one misscnse change (the latter transmitted from a father with epilepsy) in 427 ASD cases. CNVs at the *SHANKS*locus have also been identified in the genome-wide studies noted above. Altogether, these results indicate that haploinsufficicncy of *SHANKS* can cause a monogenic form of ASD with frequency of about 0.5% to 1 % of ASD cases. Furthermore, trisomy at this locus appears to result in less severe phenotypes, including Asperger syndrome and ADHD.

SHANK3 is a synaptic scaffolding protein that is abundant in the postsynaptic density (PSD). It has multiple protein interaction domains, interfacing between glutamate (and likely other) receptor complexes and actin regulatory proteins, and therefore appears to be well suited to playing a role in spine morphogenesis and synaptic plasticity. When overexpressed in cultured hippocampal neurons, SHANK3 promoted the enlargement of dendritic spines, while disruption of the related Shankl in mice led to smaller dendritic spines and reduced synaptic transmission, along with altered cognitive processes. Dramatically, expression of SHANK3 in aspiny cerebellar neurons promoted spine formation and the recruitment of glutamate receptors to the synapse, directing implicating SHANK3 in the formation and function of glutamatergic synapses.

NLGN3/4

Neuroligins (NLGNs), which are SHANK3- and NRXNl-interacting proteins, are postsynaptic CAMs that support synapse - including glutamatergic synapse - formation. There are five homologs in the human genome, with *NLGNS* and *NLGN4X* found on the Xchromosome (at Xql3 and Xp22.3, respectively), and *NLGN4Y* on the Y chromosome. Screening for mutations in these genes in over 150 cases led to the identification of two de novo mutations, including a frameshift mutation in *NLGN4* and a C-T transition in NLGN3 that led to a R451C change. In another study, a mutation in *NLGL4* that leads to a premature stop codon was found in a large pedigree. While the mutation had very high penetrance, expressivity was variable with the 10 males having mental retardation (MR) and/or ASD.*NLGN4* deletions in ASDs were observed in one recent study, but other studies have suggested that *NLGN4* deletions can be associated with little or differing psychiatric phenotypes in

some males, including tics,Tourette syndrome, and ADHD. A recent study suggests that mutations in *NGLN4Y* might also result in an ASD.

Disruption of *Nlgn4x* in mice leads to deficits in reciprocal social interactions and communication. It has been reported that introducing the R451C mutation in murine *NlgnS*was reported to result in impaired social interactions with an increase in inhibitory synaptic transmission ; however.these behavioral deficits were not seen in an independent study.

CNTNAP2

Cortical dysplasia-focal epilepsy syndrome was first described in 2006 in Amish children displaying cortical dysplasia, focal epilepsy, relative macrocephaly, diminished deep-tendon reflexes, language regression, MR, and ASD. The disorder is recessive and caused by mutations in the *CNTNAP2* gene, which codes for contactin-asso ciated protein-like 2 (CASPR2) that is involved in localization of voltage -gated potassium channels (K(v)l .1) at the juxtaparanodes of the nodes of Ran vier. Three recent studies assessed this gene in ASDs. First, following up on a linkage result of a language-related autism QTL, it was suggested that common variants of *CNTNAP2* may increase risk for ASDs in male-only families, and it was shown that *CNTNAP2* is expressed in language- and cognition-related circuits. This finding was also observed in a related study using overlapping AG RE families. Finally, rare variants of the *CNTNAP2*, and particularly the I869T variant, also show some association with ASD.

PTEN

Our own studies on *PTEN* mutations in ASD can serve to highlight the clinical value of identifying mutations in ASD. Mutations in the *PTEN* gene are associated with a broad spectrum of disorders, including Cowden syndrome (CS), Bannayan-Riley-Ruvalcaba syndrome, Proteus syndrome, and Lhermitte-Duclos disease, as well as ASDs (reviewed in ref 68). We surveyed head circumference information from hundreds of subjects with ASD and sequenced the *PTEN* gene in 88 individuals showing macrocephaly (defined as a head circumference ≥2 standard deviations above the mean). We identified a de novo missense mutation (D326N) in a highly conserved amino acid in a 5-year-old boy with autism, MR, language delay, and extreme macrocephaly (+9.6 SD).The identification of this mutation can give important information to the family with regard to recurrence risk, and also improve the care of the affected boy because an appropriate surveillance strategy for P'FfiW-mutation-related conditions can be initiated.

Etiological yield in ASDs

The importance of evaluating ASD-associated syndromes in the clinical context needs to be emphasized. A recent study used a three-tiered neurogenetic evaluation scheme to evaluate 32 patients with a behavioral diagnosis of an ASD. An overall medical and/or genetic diagnostic yield of 40% was found, although more typical studies have a lower yield (see above) and no published study has done this with modern molecular methods with an epidemiological valid cohort. As our knowledge increases and methods further evolve, it will become straightforward to carry out a comprehensive scan for genetic disorders in ASD, facilitating diagnosis, identifying medical concerns associated with syndromes, and defining subgroups that might be more responsive to specific therapeutic approaches (see below).

There is overwhelming evidence that ASDs are genetic disorders, but the genetic mechanisms are varied, involving both inherited and de novo changes, as well as mutations, trinucleotide repeats, CNVs, and larger chromosomal abnormalities. An increasing proportion of ASD is being recognized as being the result of RVs associated with high ORs.

Table I summarizes estimates the prevalence of some genetic variants in subjects ascertained for ASDs. Note that an additional 5% to 10% of cases have been identified with CNVs that are not recurrent but are likely pathogenic (based on size, de novo origin, etc). This suggests that, even with our current knowledge, 20% to 30% of ASDs can be given an etiological diagnosis using standard clinical genetic methods, including high-resolution karyotyping, array comparative genomic hybridization (array CGH), and *MECP2* sequencing in girls, as well as *PTEN* sequencing in individuals with extreme macrocephaly and the examination of methyl ation and gene dosage abnormalities in 15q.

It is of interest that synaptic and neuronal cell adhesion molecules (CAMs) are appearing in RVs in ASDs. It is also of interest that cytoplasmic proteins that bind to synaptic CAMs are also being identified. These findings

will lead to evidence-based hypotheses as to the molecular and cellular deficits underlying ASDs with differing etiologies. Of particular interest is the replicated finding of *SHANKS* deficits, which directly implicates glutamatergic synapse dysfunction in both autism and Asperger syndrome. This finding is supported by the replicated findings with *NRXN1* and *NLGNS/4,which* can also play a role in excitatory synapse formation, maintenance, and plasticity.

As the technology for detecting smaller and smaller deletions and duplications improves and as people take advantage of the newest technologies of ultradeep sequencing, the search for RVs in ASDs will enter a new phase. In this context, a useful model for the genetic and genomic architecture of ASD might be that of MR. In MR several hundred genes have been found and the evidence is strong that there are more genes to be found. Not only are some of the MR genes associated with ASDs, but as we discover more and more rare variants in autism, it is becoming increasingly clear that the architecture of MR could represent a good model as to what we will find in ASDs.

There is empirical evidence that ASD can, in some cases, respond to intensive behavioral interventions. Thus, identifying individuals with greater risks of ASD at an earlier age will have important clinical and practical implications. It will require the simultaneous analysis of multiple genetic and genomic mechanisms before effective tools for the molecular assessment of ASD etiology, used in conjunction with behavioral assessment, can be applied in a widespread manner.

As ASD loci continue to be identified, animal models that recapitulate the genetic changc(s) can be developed. These models can clarify the function of the gene products in vivo, and will ultimately be useful to evaluate novel pharmaceutical interventions. An exciting development which will serve as a useful model going forward is the elaboration of the mGluR theory of FXS. This in turn has led to the initiation of a recent large-scale clinical trial in FXS in which a reverse agonist of mGlu5 is being assessed in FXS. As additional RVs associated with ASDs are identified, novel therapeutic approaches will arise, some which may be specific to a given RV ("personalized medicine") and some that might prove effective across ASDs with differing etiologies.

Q. 3 What are the most common reasons of Attention Deficit and Hyperactivity Disorder in children? Explain the symptoms and characteristics of the disorder.

ANS: Attention deficit hyperactivity disorder (ADHD) affects children and teens and can continue into adulthood. ADHD is the most commonly diagnosed mental disorder of children. Children with ADHD may be hyperactive and unable control their impulses. Or they may have trouble paying attention. These behaviors interfere with school and home life.

It's more common in boys than in girls. It's usually discovered during the early school years, when a child begins to have problems paying attention.

Adults with ADHD may have trouble managing time, being organized, setting goals, and holding down a job. They may also have problems with relationships, self-esteem, andaddiction.

Symptoms in Children

Symptoms are grouped into three categories:

Inattention. A child with ADHD:

- Is easily distracted
- Doesn't follow directions or finish tasks
- Doesn't appear to be listening
- Doesn't pay attention and makes careless mistakes
- Forgets about daily activities
- Has problems organizing daily tasks
- Doesn't like to do things that require sitting still
- Often loses things
- Tends to daydream

Hyperactivity.

- Often squirms, fidgets, or bounces when sitting
- Doesn't stay seated
- Has trouble playing quietly
- Is always moving, such as running or climbing on things (In teens and adults, this is more commonly described as restlessness.)
- Talks excessively
- Is always "on the go" as if "driven by a motor"

Impulsivity. A child with ADHD:

- Has trouble waiting for his or her turn
- Blurts out answers
- Interrupts others

Symptoms in Adults

Symptoms of ADHD may change as a person gets older. They include:

- Chronic lateness and forgetfulness
- Anxiety
- Low self-esteem
- Problems at work
- Trouble controlling anger
- Impulsiveness
- Substance abuse or addiction
- Unorganized
- Procrastination
- Easily frustrated
- Chronic boredom
- Trouble concentrating when reading
- Mood swings
- Depression
- Relationship problems

Causes of ADHD

The cause of ADHD isn't known. Researchers say several things may lead to it, including:

- **Heredity**. ADHD tends to run in families.
- **Chemical imbalance**. Brain chemicals in people with ADHD may be out of balance.
- **Brain changes**. Areas of the brainthat control attention are less active in children with ADHD.
- **Poor nutrition, infections, smoking, drinking, and substance abuse duringpregnancy.** These things can affect a baby's brain development.
- **Toxins, such as lead.** They may affect a child's brain development.
- **A brain injury or a brain disorder.**Damage to the front of the brain, called the frontal lobe, can cause problems with controlling impulses and emotions.

Sugar doesn't cause ADHD. ADHD also isn't caused by watching too much TV, a poor home life, poor schools, or food allergies.

ADHD can't be prevented or cured. But spotting it early, plus having a good treatment and education plan, can help a child or adult with ADHD manage their symptoms.

ADHD Treatment

Many symptoms of ADHD can be managed with medication and therapy.

Medication: Medications called stimulants can help control hyperactive and impulsive behavior and increase attention span. They include:

- Dexmethylphenidate (Focalin)
- Dextroamphetamine (Adderall, Dexedrine)

- Lisdexamfetamine (Vyvanse)
- Methylphenidate (Concerta, Daytrana, Metadate, Methylin, Ritalin, Quillivant)

Stimulant medications don't work for everyone with ADHD. Nonstimulant medications may be prescribed for people older than 6. These include:

- Atomoxetine (Strattera)
- Clonidine (Kapvay)
- Guanfacine (Intuniv)

Dietary supplements with omega 3s have shown some benefit. Vayarin, a non-pharmaceutical supplement that contains omega-3s, is available by prescription only.

Therapy: These treatments focus on changing behavior.

- **Special education** helps a child learn at school. Having structure and a routine can help children with ADHD a lot.
- **Behavior modification** teaches ways to replace bad behaviors with good ones.
- **Psychotherapy (counseling**) can help someone with ADHD learn better ways to handle their emotions and frustration. It can also help improve their self-esteem. Counseling may also help family members better understand the child or adult with ADHD.
- **Social skills training** can teach behaviors, such as taking turns and sharing.

Support groups of people with similar problems and needs can help with acceptance and support. Groups also can provide a way to learn more about ADHD. These groups are helpful for adults with ADHD or parents of children with ADHD.

What to Expect

Many people with ADHD live successful, happy, full lives. Treatment helps. It's important to pay attention to symptoms and see a doctor regularly. Sometimes, medication and treatments that were once effective stop working. You may need to change the treatment plan. For many people, the symptoms of ADHD get better in early adulthood, and some are able to stop treatment

What to Expect

Many people with ADHD live successful, happy, full lives. Treatment helps. It's important to pay attention to symptoms and see a doctor regularly. Sometimes, medication and treatments that were once effective stop working. You may need to change the treatment plan. For many people, the symptoms of ADHD get better in early adulthood, and some are able to stop treatment.

Education and Training

Children and adults with ADHD need guidance and understanding from their parents, families, and teachers to reach their full potential and to succeed. Mental health professionals can educate the parents of a child with ADHD about the condition and how it affects a family. They can also help the child and his or her parents develop new skills, attitudes, and ways of relating to each other. Examples include:

- **Parenting skills training** teaches parents the skills they need to encourage and reward positive behaviors in their children.
- **Stress management techniques** can benefit parents of children with ADHD by increasing their ability to deal with frustration so that they can respond calmly to their child's behavior.
- **Support groups** can help parents and families connect with others who have similar problems and concerns.

Adding behavioral therapy, counseling, and practical support can help people with ADHD and their families to better cope with everyday problems.

School-based Programs

Some schools offer special education services to children with ADHD who qualify. Educational specialists help the child, parents, and teachers make changes to classroom and homework assignments to help the child succeed. Public schools are required to offer these services for qualified children, which may be free for families living within the school district. Learn more about the Individuals with Disabilities Education Act (IDEA),

It's your involvement that helps researchers to ultimately uncover better ways to treat, prevent, diagnose, and understand human disease. You can get involved by participating in a clinical research trial. The goal of clinical

trials is to determine if a new test or treatment works and is safe. Clinical trials can also look at other aspects of care, such as improving the quality of life for people with chronic illnesses.
Researchers at the NIMH and other NIH institutes, such as the National Human Genome Research Institute, conduct research in many areas including cognition, genetics, epidemiology, brain imaging, and treatment development. The studies take place at the NIH Clinical Center in Bethesda, Maryland. If you think you might be interested in participating in a clinical trial, you should talk to your doctor about whether to apply and identify which ones are right for you. To learn about studies on ADHD that are currently recruiting at NIMH, This is a searchable registry and results database of federally and privately supported clinical trials conducted in the United States and around the world. ClinicalTrials.gov gives you information about a trial's purpose, who may participate, locations, and phone numbers for more details. This information should be used in conjunction with advice from your health care provider.

Q. 4 Why is it necessary to carry out the assessment adaptive behaviors in children with mental retardation? What are the most commonly employed assessment techniques?

ANS:

Adaptive and Maladaptive Behavior Scales

The ***Scales of Independent Behavior - Revised*** (SIB-R), the ***Vineland Adaptive Behavior Scales***, the ***AAMR Adaptive Behavior Scales*** (ABS) and the ***Inventory for Client and Agency Planning*** (ICAP) are the most widely used adaptive behavior assessments in the United States. Their popularity is owed largely to their usefulness and accuracy, derived from quality standardization and norming. The following pages describe and compare these four tests. Unless otherwise mentioned, information comes from the tests' manuals.
Adaptive behaviors are everyday living skills such as walking, talking, getting dressed, going to school, going to work, preparing a meal, cleaning the house, etc. They are skills that a person learns in the process of adapting to his/her surroundings. Since adaptive behaviors are for the most part developmental, it is possible to describe a person's adaptive behavior as an age-equivalent score. An average five-year-old, for example, would be expected to have adaptive behavior similar to that of other five-year-olds.
Behavior problems, often called maladaptive behaviors, are behaviors that interfere with everyday activities. Good adaptive behavior and a lack of behavior problems promote independence at home, at school, and in the community. Behavior problems are much more difficult to quantify than adaptive behaviors are, because they are not very developmental and because their expression varies more from day-to-day and from setting-to-setting. Behavior problems do not increase or decrease steadily with age. Nevertheless they can be measured reliably.
The purpose of measuring adaptive and maladaptive behavior is usually either for diagnosis or for program planning. The diagnosis of mental retardation, for example, requires deficits in both cognitive ability *and* adaptive behavior, occurring before age 18. Adaptive behavior assessment is also used to determine the type and amount of special assistance that people with disabilities may need. This assistance might be in the form of home-based support services for infants and children and their families, special education and vocational training for young people, and supported work or special living arrangements such as personal care attendants, group homes, or nursing homes for adults. Adaptive behavior assessments are often used in preschool and special education programs for determining eligibility, for program planning, and for assessing outcomes.
Standardization is the process of developing a test that reliably and validly measures a specific dimension of behavior. It involves trying out items and analyzing them; revising or discarding those that don't work, adding items where there are gaps in difficulty, and reanalysis. It also involves developing standard testing procedures and materials. The result should be a test that reliably measures the same thing the same way each time, so that scores are comparable. A standardized test should also demonstrate validity, meaning that it actually measures what it portends to measure. A test score should correlate with something in the real world.

Norming is the process of determining average scores for average people. Many tests are standardized, but few are normed because norming is an expensive process which usually means testing hundreds or thousands of carefully selected individuals -- school children throughout the U.S., for example. Norms are important because they help give test scores meaning - something to compare to. A raw score of 7, for example doesn't mean much. It means more to know that a score is at the 50th percentile for an individual of a certain age.

In norming, a large number of individuals are given a standardized test in order to determine average (normal) test scores, often averages for each age or other sub-group. This permits test scores to be compared to average, to each other on a relative basis, or to scores on other normed tests. The three major types of norm scores are age scores, percentiles, and standard scores.

Tests can be normed on more narrowly defined populations, although the usefulness of these scores varies with the definition of the norm group. "Developmentally disabled adults," for example, can range from profoundly mentally retarded to almost average. Norms on such a loosely defined group are equally loosely defined. Therefore the following comparisons differentiate norms for average non-handicapped individuals from data on supplemental standardization groups of people with handicaps.

Scales Compared

This paper compares the four most widely used tests of adaptive and maladaptive behavior. Each test relies on a respondent such as a parent, teacher, or care-provider to provide information about an individual being assessed. With some tests respondents are interviewed; with other tests respondents fill out a response booklet directly.

The **Scales of Independent Behavior** (SIB-R), revised in 1996, is distinguished by several features. It contains an excellent behavior problem scale in addition to its adaptive behavior assessment, and provides a unique score which reflects overall independence based on adaptive and maladaptive behavior combined. It has norms that extend beyond adolescence -- from 3 months to over 80 years. It can be administered either as a questionnaire or as a carefully structured interview, with special materials to aid the interview process. It has a short form, a short form for children, and a short form adapted for individuals who are blind.

The **Vineland Adaptive Behavior Scales**, a revision of the original Vineland Social Maturity Scale, are distinguished by their heritage as well as by good norms and psychometrics. The Vineland assesses personal and social skills, with norms up to 18 years old. Two interview editions, one with 577 items, the other with 297, gather information through semi-structured interviews. Both include a Motor Skills Domain for children less than 6 years old and an optional Maladaptive Behavior Domain for children age 5 through 18.

The Vineland Manual states that the Survey and Expanded forms must be administered by a psychologist, social worker, or other professional with a graduate degree and training in interview techniques. Although the assessment booklets are quite straight-forward and well organized (adaptive items are ordered by difficulty and scored never, sometimes, or usually), the interview process is somewhat complex and time-consuming. The interviewer is instructed to never read items to a respondent and to never permit a respondent to read any of the items, but rather to ask general questions followed by further probes when necessary. Appendices (26 and 52 pages) describe scoring criteria for each item. A similarly administered maladaptive behavior section contains a list of 27 minor maladaptive behaviors such as *Sucks thumb or fingers*. Nine more serious behaviors, not normed for non-handicapped individuals, can be scored by frequency and noted for informational purposes as *severe* or *moderate*.

The Vineland also offers a Classroom form for children age 3 through 12. This form is a questionnaire booklet that is completed directly by a teacher. Administration of this form, which has a smaller standard error of measurement than the interview forms, requires neither interview training nor a graduate degree. The Classroom Form does not include a maladaptive behavior component.

The **AAMR Adaptive Behavior Scale 2nd edition** (AAMR ABS) was developed by the American Association on Mental Retardation, the oldest professional organization devoted to mental retardation in the United States. Its adaptive behavior scale is available in two forms -- one for School, the other Residential and Community Settings. Both versions assess the manner in which individuals cope with the natural and social demands of their environment.

The AAMR ABS adaptive behavior domains have two types of items, either "circle the highest level" or "yes/no." Some items are worded negatively, and can be somewhat confusing. A child who uses a napkin, for example, would be rated "no" on the item *Does not use a napkin*." Maladaptive behavior items are rated never, occasionally, or frequently. There

is, however, no measure of relative severity. Items such as *Blames own mistakes on others* receives the same weight as *Chokes others*.

The unidimensionality of several AAMR ABS domains is questionable. A unidimensional scale is one that measures a single dimension of knowledge or behavior, important in order to define what is being measured. A mathematics test score would be difficult or impossible to interpret if the test included a smattering of social studies questions. Likewise the AAMR Physical Development Domain score, to pick one example, is difficult to interpret because in addition to items on balance, walking, running, and arm-hand use, are items on vision and hearing, all scored together.

The **Inventory for Client and Agency Planning** (ICAP) is a 16 page booklet that, in addition to measuring adaptive and maladaptive behavior, also gathers a compact but comprehensive set of information about an individual's demographic characteristics, diagnoses, support services needed and received, and social/leisure activities. Scoring and database software prints reports and maintains current and historical information on up to 10,000 clients as well.

The ICAP's adaptive and maladaptive behavior sections contain items selected from the SIB-R, described above, with norms for infants through adults. Like the SIB-R, the ICAP also yields a Service Score, a combined measure of adaptive and maladaptive behavior indicative of overall need for care, supervision, or training. Because the ICAP is shorter than the SIB-R, its reliability is slightly less, but still excellent when compared to other scales.

Materials		
SIB-R	**Full Scale**	Manual: 287 pp. Response Booklet: 26 pp. Optional Interview Easel: 172 pp. Planning Worksheet: 2pp. (in response booklet) Software (Scoring & Reporting; PC/Mac)
	Short Form	Response Booklet: 8 pp. Response Booklet adapted for people who are blind Planning Worksheet: 2pp. Shares Full Scale Manual, Interview Easel, and software.
	Early Development Form	Response Booklet: 8 pp. Planning Worksheet: 2pp. Shares Full Scale Manual, Interview Easel, and software.
Vineland ABS	**Interview Expanded Form**	Manual: 321 pp. Item Booklet: 16 pp. Score Summary & Profile Booklet: 12pp. Program Planning Report: 8 pp. Report to Parents: 4pp. (also in Spanish) Software (Apple II/PC)
	Interview Survey Form	Manual: 301 pp. Record Booklet: 12 pp. (also in Spanish) Report to Parents: 4pp. (also in Spanish) Software (Apple II/PC)
	Classroom Edition	Manual: 175 pp. Questionnaire Booklet: 16 pp. Report to Parents: 4pp. (also in Spanish) Software (PC)

AAMR ABS 2nd Ed.	**School Edition**	Manual: 118 pp. Examination Booklet: 16pp. Profile/Scoring Form: 4 pp. Software (Scoring & Reporting; Apple/Mac/PC)
	Residential & Community Edition	Manual: 76 pp. Examination Booklet: 16pp. Profile/Scoring Form: 4 pp. Software (Scoring & Reporting; Apple/Mac/PC)
ICAP		Manual: 155 pp. Response Booklet: 16 pp. (also in Spanish) Software (Scoring & Database; PC)

SIB-R Content						
Scale subscale	**N of Items**			**Type of Score**		
	Full Scale	**Short Form**	**Early Dev.**	**Age**	**Pct**	**Std**
Broad Independence (Total)	**259**	**40**	**40**	**X**	**X**	**X**
.	.	.	.	.	.	.
Motor Skills	**38**	.	.	**X**	**X**	**X**
Gross Motor	*19*	.	.	X	.	.
Fine Motor	*19*	.	.	X	.	.
Social & Communication Skills	**56**	.	.	**X**	**X**	**X**
Social Interaction	*18*	.	.	X	.	.
Language Comprehension	*18*	.	.	X	.	.
Language Expression	*20*	.	.	X	.	.
Personal Living Skills	**88**	.	.	**X**	**X**	**X**
Eating & Meal Preparation	*19*	.	.	X	.	.
Toileting	*17*	.	.	X	.	.
Dressing	*18*	.	.	X	.	.
Personal Self-Care	*16*	.	.	X	.	.
Domestic Skills	*18*	.	.	X	.	.
Community Living Skills	**77**	.	.	**X**	**X**	**X**
Time & Punctuality	*19*	.	.	X	.	.
Money & Value	*20*	.	.	X	.	.
Work Skills	*20*	.	.	X	.	.
Home/Community Orientation	*18*	.	.	X	.	.
Maladaptive Behavior - General	**24**	**24**	**24**	.	.	**X**
.	.	.	.	.	.	.
Internalized	**9**	**9**	**9**	.	.	**X**
Hurts Self	*3*	*3*	*3*	.	.	.
Repetitive Habits	3	3	3			

Withdrawn or Inattentive	*3*	*3*	*3*	.	.	.
Asocial	**6**	**6**	**6**	.	.	**X**
Socially Offensive	*3*	*3*	*3*	.	.	.
Uncooperative	*3*	*3*	*3*	.	.	.
Externalized	**9**	**9**	**9**	.	.	**X**
Hurts Others	*3*	*3*	*3*	.	.	.
Destructive to property	*3*	*3*	*3*	.	.	.
Disruptive	*3*	*3*	*3*	.	.	.

Note. The SIB-R also provides a *Support Score*, an overall score that combines adaptive *and*maladaptive behavior.

Vineland Content

Scale subscale	N of Items			Type of Score		
	Expand	Surv	Class	Age	Pct	Std
Adaptive Behavior Composite	**541**	**261**	**244**	**X**	**X**	**X**
.	.	.	.	.	.	.
Communication	**133**	**67**	**63**	**X**	**X**	**X**
Receptive	*23*	*13*	*10*	X	.	.
Expressive	*76*	*31*	*29*	X	.	.
Written	*34*	*23*	*24*	X	.	.
Daily Living Skills	**201**	**92**	99	**X**	**X**	**X**
Personal	*90*	*39*	*36*	X	.	.
Domestic	*45*	*21*	*21*	X	.	.
Community	*66*	*32*	*42*	X	.	.
Socialization	**134**	**66**	**53**	**X**	**X**	**X**
Interpersonal Relationships	*50*	*28*	*17*	X	.	.
Play & Leisure Time	*48*	*20*	*18*	X	.	.
Coping Skills	*36*	*18*	*18*	X	.	.
Motor Skills (dev. age < 6)	**73**	**36**	**29**	**X**	**X**	**X**
Gross	*42*	*20*	*16*	X	.	.
Fine	*31*	*16*	*13*	X	.	.
Maladaptive Behavior	**36**	**36**	.	.	.	.
.	.	.	.	.	.	.
Part 1 (All children)	**27**	**27**	.	.	.	.
Part 2 (Children with handicaps)	**9**	**9**	.	.	.	.

Note. Maladaptive behavior scale yields raw scores with interpretative levels.

AAMR ABS Content

Factor/Domain	N of Items		Type of Score		
	Resid/ Cmnty	School	Age	Pct	Std
Part I (Personal independence)	**356**	**329**	.	.	.
.	.	.	.	.	.
Personal Self-Sufficiency	**103**	**103**	**X**	**X**	**X**
Independent Functioning	*76*	*76*	X	X	X
Physical Development	*27*	*27*	X	X	X
Community Self-Sufficiency	**177**	**150**	**X**	**X**	**X**
Independent Functioning	*57*	*57*	X	X	X
Economic Activity	*28*	*28*	X	X	X
Language Development	*47*	*47*	X	X	X
Numbers & Time	*15*	*15*	X	X	X
Domestic Activity	*27*	.	X	X	X
Prevocational/Vocational Activity	*3*	*3*	X	X	X
Personal-Social Responsibility	**76**	**76**	**X**	**X**	**X**
Prevocational/Vocational Activity	*9*	*9*	X	X	X
Self-Direction	*26*	*26*	X	X	X
Responsibility	*12*	*12*	X	X	X
Socialization	*29*	*29*	X	X	X
.	.	.	.	.	.
Part II (Personality/behavior)	**256**	**232**	.	.	.
.	.	.	.	.	.
Social Adjustment	**108**	**108**	.	**X**	**X**
Social Behavior	*45*	*45*	.	X	X
Conformity	*33*	*33*	.	X	X
Trustworthiness	*30*	*30*	.	X	X
Personal-Social Responsibility	**90**	**66**	.	**X**	**X**
Stereotyped & Hyperactive Behavior	*40*	*40*	.	X	X
Sexual Behavior	*24*	.	.	X	X
Self-Abusive Behavior	*26*	*26*	.	X	X
(Other)	.	.	.	.	.
Social Engagement	*23*	*23*	.	X	X
Disturbing Interpersonal Behavior	*35*	*35*	.	X	X

Note. Item types are scored *yes/no* or *select which statement best applies*. For comparability with other scales, each statement is counted as an item.

ICAP Content				
Item Scale/subscale	N of Items	Type of Score		
		Age	Pct	Std

Descriptive Characteristics	**10**	.	.	.
age/height/weight/legal status	.	.	.	.
Primary & Additional Diagnoses	**14**	.	.	.
Special Needs	**10**	.	.	.
vision/hearing/mobility	.	.	.	.
healthcare/medication	.	.	.	.
Residential Supports	**2**	.	.	.
now & in the future	.	.	.	.
School/Vocational Supports	**2**	.	.	.
now & in the future	.	.	.	.
Other Support Services	**26**	.	.	.
now & in the future	.	.	.	.
Social/Leisure Activities	**16**	.	.	.
.	.	.	.	.
Adaptive Behavior	**77**	**X**	**X**	**X**
Motor Skills	*18*	X	X	X
Social & Communication Skills	*19*	X	X	X
Personal Living Skills	*21*	X	X	X
Community Living Skills	*19*	X	X	X
.	.	.	.	.
Maladaptive Behavior	**24**	.	.	**X**
Self-injury/Stereotyped/Withdrawn	*9*	.	.	X
Offensive/Uncooperative	*6*	.	.	X
Disruptive/Destructive/Hurts others	*9*	.	.	X

Note. The ICAP also provides a *Service Score*, an overall score that combines adaptive*and* maladaptive behavior.

Standardization and Norming
Adaptive Behavior Full Scales (a)

	SIB-R	**Vineland Standard**	**AAMR School**	**ICAP**
Norm group age in yrs.	0 - 90	0 - 18 (b)	3 - 18	0 - 50
Norm group size	2,182	3,000	1,254	1,764
Supplemental standardization group (Children & adults with handicaps)	1,681	2,844	2,074 (c)	1,681
Measurement technique	Rasch	Rasch	Classic	Rasch
N of items	259	261	329	77
Standard score (SD=15) error @ 8 yrs.	±2	±4 (d)	±3	±6
Split-half/alpha reliability @ 8-9 yrs.	.98	.93	.91	.84

Test-retest reliability @ 6-13 yrs. (same interviewer 2-4 weeks apart)	.98	.85	.66 (e)	.94
Inter-rater reliability @ 6-18 yrs. (two interviewers)	.95	.74	.74 (e)	.94 (f)
Subscale intercorrelations	yes	yes	yes	yes
Construct validity - correlation with age 0-18	.91	-	.41	.91
Criterion validity - correlation with IQ (g)	.20 -.78	.28 -.52	.41 -.72	.29 -.91
Criterion validity - correlation with other AB scales	.66 -.81	.55 -.58	.53 -.61	.64 -.75
Comparison scores for age matched groups of non-handicapped students and those with hearing, learning, and emotional disabilities	yes	-	-	yes
Discriminant analysis for school placement level and level of mental retardation	yes	-	-	yes

Note. These statistics, selected from the tests' manuals, are for non-handicapped groups of comparable age, unless otherwise indicated. **Consult the tests' manuals for additional reliability and validity studies with other ages and other groups.**
(a) The AAMR does not have a total score; data are averages for the three factors The Vineland Motor Skills domain ends at age 6; data for older children are averages for 3 domains. (b) Classroom edition: age 3-12. (c) Residential & Community form: 4,103. (d) Expanded form ± 3; Classroom form ± 2. (e) Emotionally disturbed grade 9-11; no study for non-handicapped children. (f) Mentally retarded adults; no study for non-handicapped children. (g) Correlations range from high for heterogeneous groups of handicapped children to low for non-handicapped adults.

Standardization and Norming
Problem Behavior Scales

	SIB-R & ICAP (a)	**Vineland**		**AAMR School (b)**
		Part 1	**Part 2**	
Norm group age in yrs.	0 - 50	5 - 18	-	3 - 18
Norm group size	778	2,000	0	1,254
Supplemental standardization group (Children & adults with handicaps)	1,681	2,844	2,844	2,074
Development technique	Fac. anal.	-	-	Fac. anal.
N of items	16	27	9	232
Std. error of measure / SD @ 6-11 yrs.	±2.5 / 10	-	-	±3.8 / 15
Split-half/alpha reliability @ 8-9 yrs.	(c)	.87	-	.94
Test-retest reliability @ 6-13 yrs. (same interviewer 2-4 weeks apart)	.86	.88	-	.83 (d)

Inter-rater reliability @ 6-18 yrs. (two interviewers)	.83	.74	-	.57 (d)
Maladaptive subscale intercorrelations	yes	-	-	yes
Criterion validity - correlation with other maladaptive scales	.09 to .58	-	-	-
Comparison scores for age matched groups of non-handicapped students and those with hearing, learning, and emotional disabilities	yes	-	-	-
Discriminant analysis for school placement level and level of mental retardation	yes	-	-	-

Note. These statistics, selected from the tests' manuals, are for non-handicapped groups of comparable age, unless otherwise indicated. **Consult the tests' manuals for additional reliability and validity studies with other ages and other groups.** None of the four tests found consistent relationships between maladaptive behavior and intelligence. Each found a slight negative relationship between maladaptive behavior and age, and each factors age into their scoring systems.
(a) The SIB-R and the ICAP have the same problem behavior scale.
(b) The AAMR does not have a total score; data are averages for the two factors.
(c) SIB-R/ICAP maladaptive behavior categories are mutually exclusive.
(d) Emotionally disturbed grade 9-11; no study for non-handicapped children.

Q. 5 Write short note on the characteristics of the following:

- Down’s syndrome

ANS: **Down's syndrome, also known as Down syndrome or trisomy 21, is a genetic condition that typically causes some level of learning disability and certain physical characteristics.**

Characteristics of Down's syndrome

Most babies born with Down's syndrome are diagnosed soon after birth and may have:

- floppiness (hypotonia)
- eyes that slant upwards and outwards
- a small mouth with a tongue that may stick out
- a flat back of the head
- below-average weight and length at birth
- their palm may have only one crease across it

Although children with Down's syndrome share some common physical characteristics, they don't all look the same. A child with Down's will look more like their family members than other children who have the syndrome.

People with Down's syndrome will also have different personalities and abilities. Everyone born with Down's syndrome will have some degree of learning disability, but this will be different for each person.

Read more about the characteristics of Down's syndrome.

Screening for Down's syndrome

Sometimes parents find out their baby has Down's syndrome during pregnancy because of screening tests. All pregnant women are offered screening tests for Down's syndrome.

Screening tests can't tell you for certain if your baby has Down's syndrome, but they can tell you how likely it is.

If screening tests show there's a chance your baby has Down's, more tests can be done during pregnancy to confirm it.

These include:

- **chorionic villus sampling (CVS)** – a small sample of the placenta is tested, usually during weeks 11-14 of pregnancy
- **amniocentesis** – a sample of amniotic fluid is tested, usually during weeks 15-20 of pregnancy

If these tests show that your baby has Down's syndrome, you and your baby's other parent will be offered counselling so you can talk about the impact of the diagnosis.

You may also be offered an appointment to meet a doctor or other health professional who works with children with Down's syndrome. They can tell you more about the condition and answer any questions you have.

See more about screening for Down's syndrome

Causes of Down's syndrome

Down's syndrome is usually caused by an extra chromosome in a baby's cells. In most cases, this isn't inherited – it's simply the result of a one-off genetic change in the sperm or egg.

There's a small chance of having a child with Down's syndrome with any pregnancy, but the likelihood increases with the age of the mother.

For example, a woman who is 20 has about a 1 in 1,500 chance of having a baby with Down's, while a woman who is 40 has a 1 in 100 chance.

There's no evidence that anything done before or during pregnancy increases or decreases the chance of having a child with Down's syndrome.

Read more about the causes of Down's syndrome.

Living with Down's syndrome

Although there's no "cure" for Down's syndrome, there's support available to help children with the condition lead healthy, fulfilling lives.

This includes:

- access to good healthcare – including a range of different specialists
- support for your child's development – this may include speech and language therapy, physiotherapy, and home teaching
- support groups – such as the Down's Syndrome Association, who can put you in touch with other families who have a child with Down's syndrome

Lots of people with Down's syndrome are able to leave home, have relationships, work, and lead largely independent lives.

Read more about living with Down's syndrome.

Information:

Social care and support guide

If you:

- need help with day-to-day living because of illness or disability
- care for someone regularly because they're ill, elderly or disabled - including family members

Our guide to care and support explains your options and where you can get support.

Health problems linked to Down's syndrome

People with Down's syndrome are more likely to have certain health problems, including:

- heart disorders, such as congenital heart disease
- hearing and vision problems
- thyroid problems, such as an underactive thyroid gland (hypothyroidism)
- recurrent infections, such as pneumonia

Your child may be checked by a paediatrician more often than other children to pick up problems as early as possible.

If you have any concerns about your child's health, talk to your GP, health visitor or paediatrician.

- Microcephaly

ANS: **Microcephaly is a birth defect where a baby's head is smaller than expected when compared to babies of the same sex and age. Babies with microcephaly often have smaller brains that might not have developed properly.**

What is microcephaly?

Microcephaly is a condition where a baby's head is much smaller than expected. During pregnancy, a baby's head grows because the baby's brain grows. Microcephaly can occur because a baby's brain has not developed properly during pregnancy or has stopped growing after birth, which results in a smaller head size.

Microcephaly can be an isolated condition, meaning that it can occur with no other major birth defects, or it can occur in combination with other major birth defects.

What is severe microcephaly?

Severe microcephaly is a more serious, extreme form of this condition where a baby's head is much smaller than expected. Severe microcephaly can result because a baby's brain has not developed properly during pregnancy, or the brain started to develop correctly and then was damaged at some point during pregnancy.

Other Problems

Babies with microcephaly can have a range of other problems, depending on how severe their microcephaly is. Microcephaly has been linked with the following problems:

- Seizures
- Developmental delay, such as problems with speech or other developmental milestones (like sitting, standing, and walking)
- Intellectual disability (decreased ability to learn and function in daily life)
- Problems with movement and balance
- Feeding problems, such as difficulty swallowing
- Hearing loss
- Vision problems

These problems can range from mild to severe and are often lifelong. Because the baby's brain is small and underdeveloped, babies with severe microcephaly can have more of these problems, or have more difficulty with them, than babies with milder microcephaly. Severe microcephaly also can be life-threatening. Because it is difficult to predict at birth what problems a baby will have from microcephaly, babies with microcephaly often need close follow-up through regular check-ups with a healthcare provider to monitor their growth and development.

Occurrence

Microcephaly is not a common condition. State birth defects tracking systems have estimated that microcephaly ranges from 2 babies per 10,000 live births to about 12 babies per 10,000 live births in the Unites States.

Causes and Risk Factors

The causes of microcephaly in most babies are unknown. Some babies have microcephaly because of changes in their genes. Other causes of microcephaly, including severe microcephaly, can include the following exposures during pregnancy:

- Certain infections during pregnancy, such as rubella, toxoplasmosis, orcytomegalovirus
- Severe malnutrition, meaning a lack of nutrients or not getting enough food
- Exposure to harmful substances, such as alcohol, certain drugs, or toxic chemicals
- Interruption of the blood supply to the baby's brain during development

Some babies with microcephaly have been reported among mothers who were infected with Zika virus while pregnant. CDC scientists announced that enough evidence has accumulated to conclude that Zika virus infection during pregnancy is a cause of microcephaly and other severe fetal brain defects.

CDC continues to study birth defects, such as microcephaly, and how to prevent them. If you are pregnant or thinking about becoming pregnant, talk with your doctor about ways to increase your chances of having a healthy baby.

Diagnosis

Microcephaly can be diagnosed during pregnancy or after the baby is born.

During Pregnancy

During pregnancy, microcephaly can sometimes be diagnosed with an ultrasound test (which creates pictures of the body). To see microcephaly during pregnancy, the ultrasound test should be done late in the 2nd trimester or early in the third trimester. For more information about screening and confirmatory tests during pregnancy, visit CDC's birth defects diagnosis web page.

After the Baby is Born

To diagnose microcephaly after birth, a healthcare provider will measure the distance around a newborn baby's head, also called the head circumference, during a physical exam. The provider then compares this measurement to population standards by sex and age. Microcephaly is defined as a head circumference measurement that is smaller than a certain value for babies of the same age and sex. This measurement value for microcephaly is usually less than 2 standard deviations (SDs) below the average. The measurement value also may be designated as less than the 3rd percentile. This means the baby's head is extremely small compared to babies of the same age and sex.

Head circumference growth charts for newborns, infants, and children up to age 20 years in the United States can be found on CDC's growth charts website. Head circumference growth charts based on gestational age at birth (in other words, how far along the pregnancy was at the time of delivery) are also available fromINTERGROWTH 21stCdc-pdfExternal. CDC recommends that health care providers use the WHO growth charts to monitor growth for infants and children ages 0 to 2 years of age in the United States.

Microcephaly can be determined by measuring head circumference (HC) after birth. Although head circumference measurements may be influenced by molding and other factors related to delivery, the measurements should be taken on the first day of life because commonly-used birth head circumference reference charts by age and sex are based on measurements taken before 24 hours of age. The most important factor is that the head circumference is carefully measured and documented. If measurement within the first 24 hours of life is not done, the head circumference should be measured as soon as possible after birth. If the healthcare provider suspects the baby has microcephaly, he or she can request one or more tests to help confirm the diagnosis. For example, special tests like like magnetic resonance imagingcan provide critical information on the structure of the baby's brain that can help determine if the newborn baby had an infection during pregnancy. They also can help the healthcare provider look for other problems that might be present.

Webinar on Surveillance

Treatments

Microcephaly is a lifelong condition. There is no known cure or standard treatment for microcephaly. Because microcephaly can range from mild to severe, treatment options can range as well. Babies with mild microcephaly often don't experience any other problems besides small head size. These babies will need routine check-ups to monitor their growth and development.

For more severe microcephaly, babies will need care and treatment focused on managing their other health problems (mentioned above). Developmental services early in life will often help babies with microcephaly to improve and maximize their physical and intellectual abilities. These services, known as early interventionExternal, can include speech, occupational, and physical therapies. Sometimes medications also are needed to treat seizures or other symptoms.

- Hydrocephaly

ANS: The term hydrocephalus is derived from the Greek words "hydro" meaning water and "cephalus" meaning head. As the name implies, it is a condition in which the primary characteristic is excessive accumulation of fluid in the brain. Although hydrocephalus was once known as "water on the brain," the "water" is actually cerebrospinal fluid (CSF) — a clear fluid that surrounds the brain and spinal cord. The excessive accumulation of CSF results in an abnormal widening of spaces in the brain called ventricles. This widening creates potentially harmful pressure on the tissues of the brain.

The ventricular system is made up of four ventricles connected by narrow passages. Normally, CSF flows through the ventricles, exits into cisterns (closed spaces that serve as reservoirs) at the base of the brain, bathes the surfaces of the brain and spinal cord, and then reabsorbs into the bloodstream.

CSF has three important life-sustaining functions: 1) to keep the brain tissue buoyant, acting as a cushion or "shock absorber"; 2) to act as the vehicle for delivering nutrients to the brain and removing waste; and 3) to flow between the cranium and spine and compensate for changes in intracranial blood volume (the amount of blood within the brain).

The balance between production and absorption of CSF is critically important. Because CSF is made continuously, medical conditions that block its normal flow or absorption will result in an over-accumulation of CSF. The resulting pressure of the fluid against brain tissue is what causes hydrocephalus.

What are the different types of hydrocephalus?

Hydrocephalus may be congenital or acquired. Congenital hydrocephalus is present at birth and may be caused by either events or influences that occur during fetal development, or genetic abnormalities. Acquired hydrocephalus develops at the time of birth or at some point afterward. This type of hydrocephalus can affect individuals of all ages and may be caused by injury or disease.

Hydrocephalus may also be communicating or non-communicating. Communicating hydrocephalus occurs when the flow of CSF is blocked after it exits the ventricles. This form is called communicating because the CSF can still flow between the ventricles, which remain open. Non-communicating hydrocephalus — also called "obstructive" hydrocephalus — occurs when the flow of CSF is blocked along one or more of the narrow passages connecting the ventricles. One of the most common causes of hydrocephalus is "aqueductal stenosis." In this case, hydrocephalus results from a narrowing of the *aqueduct of Sylvius*, a small passage between the third and fourth ventricles in the middle of the brain.

There are two other forms of hydrocephalus which do not fit exactly into the categories mentioned above and primarily affect adults: hydrocephalus ex-vacuo and Normal Pressure Hydrocephalus (NPH).

Hydrocephalus ex-vacuo occurs when stroke or traumatic injury cause damage to the brain. In these cases, brain tissue may actually shrink. NPH is an abnormal increase of cerebrospinal fluid in the brain's ventricles that may result from a subarachnoid hemorrhage, head trauma, infection, tumor, or complications of surgery. However, many people develop NPH when none of these factors are present. An estimated 375,000 older Americans have NPH.

Who gets this disorder?

The number of people who develop hydrocephalus or who are currently living with it is difficult to establish since the condition occurs in children and adults, and can develop later in life. A 2008 data review by the University of Utah found that, in 2003, hydrocephalus accounted for 0.6 percent of all pediatric hospital admissions in the United States. Some estimates report one to two of every 1,000 babies are born with hydrocephalus.

What causes hydrocephalus?

The causes of hydrocephalus are still not well understood. Hydrocephalus may result from inherited genetic abnormalities (such as the genetic defect that causes aqueductal stenosis) or developmental disorders (such as those associated with neural tube defects including spina bifida and encephalocele). Other possible causes include complications of premature birth such as intraventricular hemorrhage, diseases such as meningitis, tumors, traumatic head injury, or subarachnoid hemorrhage, which block the exit of CSF from the ventricles to the cisterns or eliminate the passageway for CSF within the cisterns.

What are the symptoms?

Symptoms of hydrocephalus vary with age, disease progression, and individual differences in tolerance to the condition. For example, an infant's ability to compensate for increased CSF pressure and enlargement of the ventricles differs from an adult's. The infant skull can expand to accommodate the buildup of CSF because the sutures (the fibrous joints that connect the bones of the skull) have not yet closed.

In infancy, the most obvious indication of hydrocephalus is often a rapid increase in head circumference or an unusually large head size. Other symptoms may include vomiting, sleepiness, irritability, downward deviation of the eyes (also called "sun setting"), and seizures.

Older children and adults may experience different symptoms because their skulls cannot expand to accommodate the buildup of CSF. Symptoms may include headache followed by vomiting, nausea, blurred or double vision, sun setting of the eyes, problems with balance, poor coordination, gait disturbance, urinary incontinence, slowing or loss of developmental progress, lethargy, drowsiness, irritability, or other changes in personality or cognition including memory loss.

Symptoms of normal pressure hydrocephalus include problems with walking, impaired bladder control leading to urinary frequency and/or incontinence, and progressive mental impairment and dementia. An individual with this type of hydrocephalus may have a general slowing of movements or may complain that his or her feet feel "stuck." Because some of these symptoms may also be experienced in other disorders such as Alzheimer's disease, Parkinson's disease, and Creutzfeldt-Jakob disease, normal pressure hydrocephalus is often incorrectly diagnosed and never properly treated. Doctors may use a variety of tests, including brain scans such as computed tomography (CT) and magnetic resonance imaging (MRI), a spinal tap or lumbar catheter, intracranial pressure monitoring, and neuropsychological tests, to help them accurately diagnose normal pressure hydrocephalus and rule out any other conditions.

The symptoms described in this section account for the most typical ways in which progressive hydrocephalus is noticeable, but it is important to remember that symptoms vary significantly from person to person.

How is hydrocephalus diagnosed?

Hydrocephalus is diagnosed through clinical neurological evaluation and by using cranial imaging techniques such as ultrasonography, CT, MRI, or pressure-monitoring techniques. A physician selects the appropriate diagnostic tool based on an individual’s age, clinical presentation, and the presence of known or suspected abnormalities of the brain or spinal cord.

What is the current treatment?

Hydrocephalus is most often treated by surgically inserting a shunt system. This system diverts the flow of CSF from the CNS to another area of the body where it can be absorbed as part of the normal circulatory process. A shunt is a flexible but sturdy plastic tube. A shunt system consists of the shunt, a catheter, and a valve. One end of the catheter is placed within a ventricle inside the brain or in the CSF outside the spinal cord. The other end of the catheter is commonly placed within the abdominal cavity, but may also be placed at other sites in the body such as a chamber of the heart or areas around the lung where the CSF can drain and be absorbed. A valve located along the catheter maintains one-way flow and regulates the rate of CSF flow.

A limited number of individuals can be treated with an alternative procedure called third ventriculostomy. In this procedure, a neuroendoscope — a small camera that uses fiber optic technology to visualize small and difficult to reach surgical areas — allows a doctor to view the ventricular surface. Once the scope is guided into position, a small tool makes a tiny hole in the floor of the third ventricle, which allows the CSF to bypass the obstruction and flow toward the site of resorption around the surface of the brain.

What are the possible complications of a shunt system?

Shunt systems are imperfect devices. Complications may include mechanical failure, infections, obstructions, and the need to lengthen or replace the catheter. Generally, shunt systems require monitoring and regular medical follow up. When complications occur, subsequent surgery to replace the failed part or the entire shunt system may be needed.

Some complications can lead to other problems such as overdraining or underdraining. Overdraining occurs when the shunt allows CSF to drain from the ventricles more quickly than it is produced. Overdraining can cause the ventricles to collapse, tearing blood vessels and causing headache, hemorrhage (subdural hematoma), or slit-like ventricles (slit ventricle syndrome). Underdraining occurs when CSF is not removed quickly enough and the symptoms of hydrocephalus recur. Overdrainage and underdrainage of CSF are addressed by adjusting the drainage pressure of the shunt valve; if the shunt has an adjustable pressure valve these changes can be made by placing a special magnet on the scalp over the valve. In addition to the common symptoms of hydrocephalus, infections from a shunt may also produce symptoms such as a low-grade fever, soreness of the neck or shoulder muscles, and redness or tenderness along the shunt tract. When there is reason to suspect that a shunt system is not functioning properly (for example, if the symptoms of hydrocephalus return), medical attention should be sought immediately.

What is the prognosis?

The prognosis for individuals diagnosed with hydrocephalus is difficult to predict, although there is some correlation between the specific cause of the hydrocephalus and the outcome. Prognosis is further clouded by the presence of associated disorders, the timeliness of diagnosis, and the success of treatment. The degree to which relief of CSF pressure following shunt surgery can minimize or reverse damage to the brain is not well understood.

Affected individuals and their families should be aware that hydrocephalus poses risks to both cognitive and physical development. However, many children diagnosed with the disorder benefit from rehabilitation therapies and educational interventions and go on to lead normal lives with few limitations. Treatment by an interdisciplinary team of medical professionals, rehabilitation specialists, and educational experts is critical to a positive outcome. Left untreated, progressive hydrocephalus may be fatal.

The symptoms of normal pressure hydrocephalus usually get worse over time if the condition is not treated, although some people may experience temporary improvements. While the success of treatment with shunts varies from person to person, some people recover almost completely after treatment and have a good quality of life. Early diagnosis and treatment improves the chance of a good recovery.

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What research is being done?

The National Institute of Neurological Disorders and Stroke (NINDS) and other institutes of the National Institutes of Health (NIH) conduct research related to hydrocephalus and support additional research through grants to major medical research institutions across the country. Much of this research focuses on finding better ways to prevent, treat, and ultimately cure disorders such as hydrocephalus. The NINDS also conducts and supports a wide range of fundamental studies that explore the complex mechanisms of normal and abnormal brain development.

The Hydrocephalus Clinical Research Network (HCRN, ***www.hcrn.org)*** is a multi-center collaborative research effort that was borne out of the first NIH workshop on hydrocephalus. NINDS supported the work of HCRN through the Challenge Grant process to advance their studies. HCRN consists of seven pediatric centers that pool their hydrocephalus patient population to more rapidly study the potential for improved treatments. HCRN conducts multiple, simultaneous studies at all of its centers and maintains a substantial registry of patients and procedures.

Mental Disorder

ANS: Mental illness refers to a wide range of mental health conditions — disorders that affect your mood, thinking and behavior. Examples of mental illness include depression, anxiety disorders, schizophrenia, eating disorders and addictive behaviors.

Many people have mental health concerns from time to time. But a mental health concern becomes a mental illness when ongoing signs and symptoms cause frequent stress and affect your ability to function.

A mental illness can make you miserable and can cause problems in your daily life, such as at school or work or in relationships. In most cases, symptoms can be managed with a combination of medications and talk therapy (psychotherapy).

Symptoms

Signs and symptoms of mental illness can vary, depending on the disorder, circumstances and other factors. Mental illness symptoms can affect emotions, thoughts and behaviors.

Examples of signs and symptoms include:

- Feeling sad or down
- Confused thinking or reduced ability to concentrate
- Excessive fears or worries, or extreme feelings of guilt
- Extreme mood changes of highs and lows
- Withdrawal from friends and activities

- Significant tiredness, low energy or problems sleeping
- Detachment from reality (delusions), paranoia or hallucinations
- Inability to cope with daily problems or stress
- Trouble understanding and relating to situations and to people
- Alcohol or drug abuse
- Major changes in eating habits
- Sex drive changes
- Excessive anger, hostility or violence
- Suicidal thinking

Sometimes symptoms of a mental health disorder appear as physical problems, such as stomach pain, back pain, headache, or other unexplained aches and pains.

When to see a doctor

If you have any signs or symptoms of a mental illness, see your primary care provider or mental health specialist. Most mental illnesses don't improve on their own, and if untreated, a mental illness may get worse over time and cause serious problems.

If you have suicidal thoughts

Suicidal thoughts and behavior are common with some mental illnesses. If you think you may hurt yourself or attempt suicide, get help right away:

- Call 911 or your local emergency number immediately.
- Call your mental health specialist.
- Call a suicide hotline number — in the U.S., call the National Suicide Prevention Lifeline at 1-800-273-TALK (1-800-273-8255).
- Seek help from your primary care doctor or other health care provider.
- Reach out to a close friend or loved one.
- Contact a minister, spiritual leader or someone else in your faith community.

Suicidal thinking doesn't get better on its own — so get help.

Helping a loved one

If your loved one shows signs of mental illness, have an open and honest discussion with him or her about your concerns. You may not be able to force someone to get professional care, but you can offer encouragement and support. You can also help your loved one find a qualified mental health provider and make an appointment. You may even be able to go along to the appointment.

If your loved one has done self-harm or is considering doing so, take the person to the hospital or call for emergency help.

Request an Appointment at Mayo Clinic

Causes

Mental illnesses, in general, are thought to be caused by a variety of genetic and environmental factors:

- **Inherited traits.** Mental illness is more common in people whose blood relatives also have a mental illness. Certain genes may increase your risk of developing a mental illness, and your life situation may trigger it.
- **Environmental exposures before birth.** Exposure to environmental stressors, inflammatory conditions, toxins, alcohol or drugs while in the womb can sometimes be linked to mental illness.
- **Brain chemistry.** Neurotransmitters are naturally occurring brain chemicals that carry signals to other parts of your brain and body. When the neural networks involving these chemicals are impaired, the function of nerve receptors and nerve systems change, leading to depression.

Risk factors

Certain factors may increase your risk of developing mental health problems, including:

- Having a blood relative, such as a parent or sibling, with a mental illness
- Stressful life situations, such as financial problems, a loved one's death or a divorce
- An ongoing (chronic) medical condition, such as diabetes
- Brain damage as a result of a serious injury (traumatic brain injury), such as a violent blow to the head
- Traumatic experiences, such as military combat or being assaulted

- Use of alcohol or recreational drugs
- Being abused or neglected as a child
- Having few friends or few healthy relationships
- A previous mental illness

Mental illness is common. About 1 in 5 adults has a mental illness in any given year. Mental illness can begin at any age, from childhood through later adult years, but most begin earlier in life.

The effects of mental illness can be temporary or long lasting. You also can have more than one mental health disorder at the same time. For example, you may have depression and a substance use disorder.

Complications

Mental illness is a leading cause of disability. Untreated mental illness can cause severe emotional, behavioral and physical health problems. Complications sometimes linked to mental illness include:

- Unhappiness and decreased enjoyment of life
- Family conflicts
- Relationship difficulties
- Social isolation
- Problems with tobacco, alcohol and other drugs
- Missed work or school, or other problems related to work or school
- Legal and financial problems
- Poverty and homelessness
- Self-harm and harm to others, including suicide or homicide
- Weakened immune system, so your body has a hard time resisting infections
- Heart disease and other medical conditions

Prevention

There's no sure way to prevent mental illness. However, if you have a mental illness, taking steps to control stress, to increase your resilience and to boost low self-esteem may help keep your symptoms under control. Follow these steps:

- **Pay attention to warning signs.** Work with your doctor or therapist to learn what might trigger your symptoms. Make a plan so that you know what to do if symptoms return. Contact your doctor or therapist if you notice any changes in symptoms or how you feel. Consider involving family members or friends to watch for warning signs.
- **Get routine medical care.** Don't neglect checkups or skip visits to your health care provider, especially if you aren't feeling well. You may have a new health problem that needs to be treated, or you may be experiencing side effects of medication.
- **Get help when you need it.** Mental health conditions can be harder to treat if you wait until symptoms get bad. Long-term maintenance treatment also may help prevent a relapse of symptoms.
- **Take good care of yourself.** Sufficient sleep, healthy eating and regular physical activity are important. Try to maintain a regular schedule. Talk to your health care provider if you have trouble sleeping or if you have questions about diet and physical activity.