UWJ cc Appr 3/21/06
05-45 Senate Info 5/2/06

TO Provost 3/22/06

Undergraduate Distance Education Review Form

(Required for all courses taught by distance education for more than one-third of teaching contact hours.)

Existing and Special Topics Course

Received
FEB 22 2006
Liberal Studies

Course: 418

Instructor(s) of Record: Dr. Joseph Domaracki

Phone: 7-2456 **Email:** jwdomara@iup.edu

Step One: Proposer

A. Provide a brief narrative rationale for each of the items, A1- A5.

1. How is/are the instructor(s) qualified in the distance education delivery method as well as the discipline?
2. How will each objective in the course be met using distance education technologies?
3. How will instructor-student and student-student, if applicable, interaction take place?
4. How will student achievement be evaluated?
5. How will academic honesty for tests and assignments be addressed?

B. Submit to the department or its curriculum committee the responses to items A1-A5, the current official syllabus of record, along with the instructor developed online version of the syllabus, and the sample lesson. This lesson should clearly demonstrate how the distance education instructional format adequately assists students to meet a course objective(s) using online or distance technology. It should relate to one concrete topic area indicated on the syllabus.

Step Two: Departmental/Dean Approval

Recommendation: ☒ Positive (The objectives of this course can be met via distance education)

☐ Negative

Joseph Domaracki 2.20.06
Signature of Department Designee Date

Endorsed: Mary Ann Rafoth 2.21.06
Signature of College Dean Date

Forward form and supporting materials to Liberal Studies Office for consideration by the University-wide Undergraduate Curriculum Committee. Dual-level courses also require review by the University-wide Graduate Committee for graduate-level section.

Step Three: University-wide Undergraduate Curriculum Committee Approval

Recommendation: ☒ Positive (The objectives of this course can be met via distance education)

☐ Negative

Gail S Sechrist 3-21-06

Signature of Committee Co-Chair Date

Forward form and supporting materials to the Provost within 30 calendar days after received by committee.

Step Four: Provost Approval

☐ Approved as distance education course ☐ Rejected as distance education course

Signature of Provost Date

Forward form and supporting materials to Associate Provost.

Undergraduate Distance Education Review Form

Step One: Proposer

A. Provide a brief narrative rationale for each of the items, A1-A5

1. How is/are the instructor(s) qualified in the distance education delivery mode as well as the discipline?

   The instructor of this course holds a Ph.D. in Special Education with a specialization in Severe Disabilities and has taught this course in the classroom twice annually from January 1992 through May of 2002 and again in the fall of 2005 and the spring of 2006. The instructor has been using WebCT as an instructional tool for grading purposes in EDEX 435 and EDEX 418 for the past two academic years and during that time has learned how to utilize WebCT to build and implement this course.

2. How will each objective in the course be met using distance technologies?

   The same content and assignments used in the classroom version of this course, which address the objectives of the course, has been incorporated into Instructional Modules available to students through WebCT. Each Instructional Module is designed to include a PowerPoint slide presentation containing links to websites that present students with videos, illustrations and readings related to the topics addressed by course objectives.

3. How will instructor –student and student–student, if applicable, interaction take place?

   The course is designed with a discussion room. Each module has an associated discussion question designed to facilitate student-student and instructor –student interaction. The instructor monitors discussion entries regularly and provides feedback to comments made by students as well as posing follow-up questions to stimulate further discussion. There is a rubric associated with the discussion room questions. Students will earn participation points based on the number and quality of entries submitted to the discussion. Additionally, there is a built in e-mail component to the course as well so that all interaction relevant to the course, and not related to discussion questions, can be done through the on-line course and not regular university or outside ISP networks.

4. How will student achievement be evaluated?

   The course has fourteen (14) Instructional Modules. Each Instructional Module has a quiz and a discussion question associated with it. Students will be evaluated based on quiz performance and discussion participation. Additionally, students will be required to electronically submit one six (6) page website review paper on a topic relevant to the course. Course topics must be approved by the instructor.

5. How will academic honesty for tests and assignments be addressed?

   Quizzes associated with the Instructional Modules have been designed using Respondus. Quizzes are time released and once closed will not be opened again. When the quizzes are open students will have ten minutes to respond to all questions. Students make take each quiz two (2) times. Website review topics are controlled by the instructor to prevent duplication thus avoiding the potential for dishonesty.

COURSE SYLLABUS

I. CATALOG DESCRIPTION

EX 418/518 EDUCATION OF PERSONS WITH PHYSICAL OR MULTIPLE DISABILITIES
3 s.h.

Focuses on major theoretical positions regarding etiology of a wide and diverse range of physical/multiple disabilities, the definition and identification of the population, and educational approaches. Reviews research in the field, including current issues, trends, practices, and services.

II. COURSE OBJECTIVES

This course is designed to develop:

1. knowledge and understanding of definitions, terminology, and the identification process;

2. knowledge and understanding of characteristics of physically or multiply disabled children, adolescents, and adults: educational and emotional needs, motivational and learning characteristics, and vocational and career development needs;

3. knowledge and understanding of federal and state legislation related to service delivery and program development, including PL 94-142, PL 99-457, PL 101-476, Section 504, and the Americans with Disabilities Act;

4. knowledge and understanding of research and theoretical orientations regarding etiologies and interventions;

5. knowledge and understanding of current issues and trends;

6. knowledge and understanding of the relevance of multicultural and linguistic diversity in identification and service delivery;

7. knowledge and understanding of appropriate educational programming in the following service delivery models: instructional support, itinerant, resource, self-contained, and transition;

8. knowledge and understanding of therapeutic management of sensorimotor and physical disabilities;

9. knowledge and understanding of avenues to physical participation;

10. knowledge and understanding of collaborative consultation with professionals and parents;

11. knowledge and skill in using computer software, alternate programming, and current technology;

12. skill in applying formal and informal assessment data to guide instructional decision-making.

III. COURSE OUTLINE

A. Definitions and Terminology

B. Characteristics of Conditions Resulting in Physical Disabilities

C. Historical Perspectives

D. Service Delivery Models

1. Integrated placement model
2. Segregated placement model

E. Theoretical Perspectives: Concepts and Implications

1. Classification, etiological and descriptive research, and educational approaches
2. Integration issues

G. Medical Aspects

H. Research and Issues in Assessment Procedures

1. Types of measures
2. Educational relevance of assessment measures
3. Technical adequacy and usefulness
4. Biasing factors, including concerns regarding students from multiculturally and linguistically diverse backgrounds

I. Learning Characteristics: Preschool through Adult

1. Academic skills
2. Social and emotional aspects
3. Career/vocational implications
4. Special health care needs

J. Principles of Academic Remediation and Program effectiveness

1. Prescriptive teaching
2. Life management skills
3. Communication skills
4. Physical and motor skills
5. Linguistically and culturally diverse students
6. Use of technology to enhance instruction

K. Collaborative Consultation and Teamwork

1. General educators and other agency personnel
2. Parents

IV. PRE-PRACTICUM FIELD EXPERIENCES FOR GRADUATE STUDENTS

The graduate-level version of this course is designed primarily for students seeking post-baccalaureate certification.

The graduate student is required to spend a total of 20 hours of participation in a setting serving students with physical and/or multiple disabilities. The field experience may be completed prior to or concurrent with course enrollment, as long as all participation hours are completed by the midterm point in the course. Students are expected to provide written and oral accounts of their experiences according to instructor guidelines and departmental policy governing pre-student teaching experiences.

V. COURSE REQUIREMENTS

Students will demonstrate mastery of course objectives through successful completion of the following course requirements:

Completion of assigned readings in text and selected journals.

Participation in class discussion and/or activities.

Exams. Three major exams consisting of multiple choice, completion and short essay items. 100 points.

Course Project. Each student will design a specific instructional program or adaptive skill program that could be used in educating an individual with physical or multiple disability. The plan should include: objective/purpose, procedures for use of the plan, application of the plan and methods for evaluating the plans effectiveness. 100 points.

Journal Critiques. Each student will complete critiques/analysis of five articles from professional journals. The critique must address a variety of topics discussed during the course (no two articles can address the same topic). Each critique/analysis must be 2-3 pages in length. 20 points each (100 points total).

GRADUATE STUDENTS ONLY:
Field Experience. See item IV Pre-Practicum Field Experience for Graduate Students for detailed description of assignment.

Literature Review Paper. Each student will complete a 5-7 page literature review paper on a topic directly related to course content. The paper must include at least five references that are no more than 5 years old. Topics must be approved by the course instructor.

VI. EVALUATION METHODS

The course grade will be interpreted from the total accumulated points

and weighted according to the following scale:

		Undergraduate	Graduate
A = 90-100%	Exams	50%	25%
B = 80-89%	Course Project	25%	15%
C = 70-79%	Journal Critique	25%	10%
D = 60-69%	Field Experience		25%
F = 59% or below	Literature Review		25%

*No "D" grade is recognized in IUP graduate work.

VII. REQUIRED TEXTS

Bigge, J. L. (1991). Teaching individuals with physical and multiple disabilities. New York: Merrill.

VIII. BIBLIOGRAPHY

Batshaw, M. L., & Perret, Y. M. (1986). Children with handicaps: A medical primer. Baltimore: Paul Brookes.

Bleck, E. E., & Nagel, D. A. (1982). Physically handicapped children: A medical atlas for teachers. Boston: Allyn & Bacon.

Church, G., & Glennen, S. (1991). The handbook of assistive technology. San Diego: Singular Publishing.

Fraser, B. A., & Hensinger, R. (1983). Managing physical handicaps. Baltimore: Paul H. Brookes.

Goossens, C., & Crain, S. (1986). Augmentative communication: Intervention resource. Wauconda, IL: Don Johnston Developmental Equipment.

Hardy, J. C. (1983). Cerebral Palsy. Englewood Cliffs, NJ: Prentice Hall.

Henderson, G., & Bryan, W. V. (1984). Psychosocial aspects of disability. Springfield: Charles C. Thomas.

Hilgartner, M. (1989). Hemophilia in the child and adult. New York: Raven Press.

Holliday, P. (1989). Special needs in ordinary schools: Children with physical disabilities. London: Carrell Education Ltd.

Holvoet, J., & Helmstetter, E. (1989). Medical problems of students with special needs: A guide for educators. Boston: College Hill.

McLone, D. (1984). An introduction to spina bifida. Chicago: Northwestern University.

Setoguchi, Y., & Rosenfelder, R. (1982). The limb deficient child. Springfield, IL: Charles C. Thomas.

Sowers, J. A., & Powers, L. (1991). Vocational preparation and employment of students with physical and multiple disabilities. Baltimore, MD: Paul H. Brookes.

Tver, D. F., & Tver, B. M. (1991). Encyclopedia of mental and physical handicaps. Austin, TX: Pro-Ed.

Umbreit, J. (1983). Physical disabilities and health impairments: An introduction. Columbus, Merrill.

Urbano, M. T. (1992). Preschool children with special health care needs. San Diego: Singular Publishing.

Wehman, P., Wood, W., Everson, J. M., Goodwin, R., & Conley, S. (1988). Vocational education for multihandicapped youth with cerebral palsy. Baltimore, MD: Paul H. Brookes Publishing Co.

Williamson, G. G. (1987). Children with spina bifida: Early intervention and preschool programming. Baltimore: Paul H. Brookes.

5/93

COURSE TITLE: EDEX. 418 / 518 - Education of Persons with Physical and Multiple Disabilities

TERM: Spring Semester 2006 (05-50)

LOCATION: Web -CT

INSTRUCTOR: Joseph Domaracki, Ph.D.

OFFICE: 215 Davis Hall
357-2456
Hours: Mondays – 11:30 – 1:00
Wednesdays - 10:00 – 12:00
Fridays - 11:30 – 1:00
Other times by appointment.

COURSE OBJECTIVES: This course is designed to develop:

A. knowledge and understanding of definitions, terminology, and the identification process; **Chapter 354: I.A, I.C; CEC: Standard 1 & 2**

B. knowledge and understanding of characteristics of physically or multiply disabled children, adolescents, and adults: educational and emotional needs, motivational and learning characteristics, and vocational and career development needs; **Chapter 354: I.B, I.D; CEC: Standard 2 & 3**

C. knowledge and understanding of federal and state legislation related to service delivery and program development, including PL 94-142, PL 99-457, PL 101-476, Section 504, and the Americans with Disabilities Act; **Chapter 354: I.A, I.C, I.F; CEC: Standard 1 & 2**

D. knowledge and understanding of research and theoretical orientations regarding etiologies and interventions; **Chapter 354: I.C, I.D, I.E; CEC: Standard 2 & 3**

E. knowledge and understanding of current issues and trends; **Chapter 354: I.A, I.B, I.C, I.F;III.A; CEC: Standard 1, 2 & 9**

F. knowledge and understanding of the relevance of multicultural and linguistic diversity in identification and service delivery; **Chapter 354: I.B, I.F; CEC: Standard 1 & 2**

G. knowledge and understanding of appropriate educational programming in the following service delivery models: instructional support, itinerant, resource, self-contained, and transition; **Chapter 354: I.G, I.I, I.J; II.A, II.B, II.C, II.D, II.F; CEC: Standard 4, 5, 6, 7, & 8**

H. knowledge and understanding of therapeutic management of sensorimotor and physical disabilities; **Chapter 354: I.C, I.D, I.E; II.D; CEC: Standard 2, 3, 5 & 7**

I. knowledge and understanding of avenues to physical participation; **Chapter 354: II.B, II.D; CEC: Standard 5 & 7**

J. knowledge and understanding of collaborative consultation with professionals and parents; **Chapter 354: II.A, II.G; CEC: Standard 4**

K. knowledge and skill in using computer software, alternate programming, and current technology; **Chapter 354: I.J; CEC: Standard 4**

L. skill in applying formal and informal assessment data to guide instructional decision-making. **Chapter 354: I.G, I.H, I.I, I.J; II.A, II.G; CEC: Standard 4, 6, 7 & 8**

Discipline Specific Standard/ Program Objective	Course Objective and Performance Indicator	Course Assessment Measuring Objective
1. Foundation	A, C, E, & F	Web Site Review Paper Literature Review Paper Exam 1
2. Development & Characteristics	A, B, C, D, E, & F	Web Site Review Paper Literature Review Paper Exams 1, 2, & 3
3. Learning Differences	B & D	Web Site Review Paper Literature Review Paper Exams 1, 2, & 3
4. Instructional Strategies	G, H, I, J, K, &L	Web Site Review Paper Literature Review Paper Exam 2
5. Environmental & Social Interaction	F, G, I,	Web Site Review Paper Literature Review Paper Exams 1, 2 & 3
6. Communication	B, D, F, G, I, J, K &L	Web Site Review Paper Literature Review Paper Exam 2
7. Instructional Planning	G, J &L	Web Site Review Paper Literature Review Paper Exam 2
8. Assessment	G &L	Web Site Review Paper Literature Review Paper Exam 3
9. Ethics	C & G	Web Site Review Paper Literature Review Paper Exam 3
10. Collaboration	E & J	Web Site Review Paper Literature Review Paper

REQUIREMENTS:

1). Complete course readings.
2). Participate in class discussion and activities.
3). Complete course exams.
4). Attend all class meetings
5). Pre-practicum field experience observations - **Graduate Students Only**
6). Complete Literature Review Paper - **Graduate Students Only**

PRE-PRACTICUM FIELD EXPERIENCE:

The graduate-level version of this course is designed primarily for students seeking post-baccalaureate certification. Each student is required to spend a total of 20 hours of participation in a setting that serves students with mental retardation or developmental disabilities. The field experience may be completed prior to or concurrent with course enrollment. Students are expected to provide a written account of their experience. The written assignment for this experience will be a daily journal chronicling the 20-hour observation experience. Evaluation and grading of this journal is explained in the "Grades" section of the syllabus.

COURSE QUIZZES

QUIZZES - There will be one quiz for each module of the course. If you score less than 80% on a quiz you will be allowed one retake to achieve an 80 % or better.

WRITTEN ASSIGNMENTS:

Graduate students are required to write a Literature Review Paper on a topic of choice related to this course. The literature review paper should be 5 – 7 pages in length and should have a minimum of seven (7) recent (within the last seven years) references. References may include web sites, journal articles and books.

Graduate and Undergraduate students are required to complete a Web Site Resource Review Paper on a topic of choice related to this course. This document is to be 7 pages in length and should review three (3) reputable web sites. The seven pages will consist of a title page (pg 1), an introduction page (pg 2), a one (1) page review per web site including web address pgs 3-5), a conclusion page (pg 6 and a reference list (pg 7). Missing or extra pages will result in a two point deduction. This assignment must be typed, double-spaced and written in APA format and submitted via e-mail as an attachment.

Written assignments will be evaluated and graded using the following criteria:

a) Sentence Structure - complete and grammatically correct;
b) Word usage - appropriate form, tense, and person;
c) Punctuation and spelling - correct usage and spelling;
d) Terminology - appropriate use of professional terms; and
e) Overall organization of content.

All written assignments must be typed using Microsoft Office Suite. Any assignment that is not typed will not be accepted. Additionally, all assignments must be turned in ON TIME!!! If by chance your assignment is late there will be a five (5)-point deduction from your grade. Late assignments will only be accepted for two (2) days following the due date, any assignment that is more than two days late WILL NOT, I repeat, WILL NOT be accepted except in cases of illness verified by a doctors excuse.

DICUSSION ENTRIES

This course is designed with a discussion room. For each topic there will be discussion question available each week. There is no correct or incorrect manner in which to answer/address each question. Rather, the questions are designed to stimulate ideas related to the question or issue at hand. All students are expected to submit discussion entries for each week's question. In order to guide discussion there are several rules that will govern operation of the discussion room and a rubric for earning points. They rules and rubric are as follows:

RULES

1. All entries must be submitted in complete, well structured and grammatically correct sentences.
2. An entry may not be one in which you solely agree with a previous comment. You are allowed to agree, but, you must also extend the discussion with additional thoughts of your own. This will help facilitate a threaded discussion (see directions on discussion page)
3. There is no right or wrong thought/opinion. All thoughts/opinions may be expressed as long as the thought/opinion is stated tastefully avoiding foul language, ethnic/racial slurs or disability insensitive language. Of course, all of the previously stated requirements for foul language, slurs and insensitivity are judgment calls. I will be the judge of what is foul, slurring or insensitive. If you are found in violation of using foul, slurring or insensitive language your entry will denied and not counted toward earned points. All rulings are final and not subject to "red flag review".

RUBRIC

Each student can earn up to ten (10) points per discussion question. Because the objective

of the discussion room is to facilitate group interaction via threaded discussion students are expected to log more than one (1) or two (2) entries. Therefore, the rubric for awarding points is as follows:

one (1) – three (3) entries are worth (3) points
three (3) – six (6) entries are worth six (6) points
seven (7) plus entries are worth ten (10) points

GRADES: Points will be accumulated during the semester from the quizzes and written assignments. Points will be distributed in the following manner.

Quizzes –140 points (10 points per module)
Literature Review Paper – 100 points
Pre-practicum Paper 100 points
Web Site Resource Review –120 points
Discussion Entry – 140 points (10 per discussion question)

		UNGRAD	GRADUATE
93 - 100%	A	369 - 400	555 - 600
85 - 92%	B	338 - 368	507 - 554
77 - 84%	C	306 - 337	459 - 506
69 - 76%	D	274 - 305	411 - 458
0 - 68%	F	273 & Below	410 & Below

Course Module 1

Conditions Resulting in Physical Disabilities

N.B. – Underlined terms within the bulleted portions of the Power Point presentation are linked to websites. The linked websites are incorporated to do one of three things:

1. Provide students with a more detailed written description about the topic/concept/process.

2. Provide an illustration (single image or series of images) of the topic/concept/process.

3. Provide a short video that illustrates the topic/concept/process.

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

CHROMOSOMES

- Each cell has 23 complimentary pair of chromosomes (46 chromosomes in total).
- Each parent contributes a set of 23 chromosomes.
- Chromosomes direct all cellular activities.

+ 32 more slides

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

CHROMOSOMES

- Exceptions to the rule are the germ cells.
- Germ cells are the female's egg and the male's sperm.
- Each germ cell only has 23 chromosomes and must join with another germ cell to create 23 complimentary pair.

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

CHROMOSOMES

- Pairs 1 through 22 are complimentary sets of chromosomes and contain identical genetic information necessary to sustain life.
- Pairs 1 - 22 are called Autosomes.
- The 23rd pair are called Sex Chromosomes and contain the genetic material that makes us different from our siblings.

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

CELL DIVISION

- Mitosis is the process of cell division that all cells in our body experience. It consists of four phases:
  - Prophase
  - Metaphase
  - Anaphase
  - Telophase

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

CELL DIVISION

- **Germ cells are different. Germ cells experience a process called Meiosis.**
- **Meiosis contains the same phases as Mitosis with one exception.**
- **The exception is that in Meiosis each phase occurs twice.**

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

CELL DIVISION

- The Meiotic process involves a primary spermatocyte or oocyte which both contain 46 chromosomes.

- The primary spermatocytes and oocytes are produced in the testes of the male and the ovaries of the female.

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

CELL DIVISION

- The primary spermatocyte or oocyte initially experience a modified Mitotic process of the four phases in which a reductive division occurs.

- The primary spermatocytes and oocytes are reduced from one cell containing 46 chromosomes to two cells containing 23 chromosomes. These two cells are called secondary spermatocytes and oocytes.

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

CELL DIVISION

- The secondary spermatocyte or oocyte will now experience a second round of the four phases of Mitosis in which a reproductive division occurs.

- The two secondary spermatocytes and oocytes are now reproduced to create four cells (sperm and ovum) containing 23 chromosomes.

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

PROBLEMS IN CELL DIVISION

- Nondisjunction – The unequal division of chromosomes in the Telophase stage of Mitosis.
  - Instead of splitting 1:1, 2 chromosomes go to one cell and the other cell gets nothing.
  - Results in a 22-24 split v. 23-23 split
  - More frequent in the egg(85%) v. the sperm (15%)

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

PROBLEMS IN CELL DIVISION

- If fertilized the cell receiving 24 chromosomes can survive but the cell receiving 23 chromosomes will not.
- This problem occurs most frequently with the 21st chromosome.
- If a cell with 24 chromosomes (2 #21) is fertilized then a Trisomy 21 ocurs. This is Down Syndrome.

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

PROBLEMS IN CELL DIVISION

- Turner Syndrome is another nondisjunction problem.
- This problem occurs with the X chromosome v. other chromosomes.
- In this syndrome a female is born with only one X chromosome. This is the only known syndrome in which someone can survive with 45 chromosomes.

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

PROBLEMS IN CELL DIVISION

- Deletion-The breaking off of a piece of a chromosome during the Telophase stage of Mitosis.
  - The detached portion of the chromosome is lost or may attach to another chromosome.
  - Cri-du-Chat Syndrome is an example of deletion involving a lost section of the #5 chromosome.

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

PROBLEMS IN CELL DIVISION

- Translocation-The transfer of a piece of a deleted chromosome to another chromosome.
  - This also frequently happens with the #21 chromosome. A part of a #21 may attach to a #14. This creates a cell with 1 #21 and 1 #14/21 combination. If fertilized the new cell will have 46 chromosomes with 2 #21s, 1 #14 and 1 #14/21 combination.

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

PROBLEMS IN CELL DIVISION

- The result of this type of translocation is a partial Trisomy 21.
- Partial Trisomy 21 is another version of Down Syndrome

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

PROBLEMS IN CELL DIVISION

- Most conceptions with chromosomal abnormalities spontaneously abort.
- It is estimated that over 50% of all miscarriages prior to 12 weeks are chromosomal abnormalities.
- After 12 weeks the frequency of spontaneous abortion due chromosomal abnormality is greatly decreased.

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

HEREDITY

- Many disabilities are inherited and can be classified in several ways. They can be classified as a:
  - Autosomal Recessive Disorder
  - Autosomal Dominant Disorder
  - Sex Linked/X Linked Disorder

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

HEREDITY

- Autosomal Recessive Disorders
  - Can only occur if two persons carry the gene for a particular disorder.
  - Affect males and females equally.
  - Results in enzymatic/biochemical abnormalities.
  - Is the result of a history of intermarriage.

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

HEREDITY

- Autosomal Recessive Disorders
  - Tay–Sack Disease
  - Galactosemia
  - Maple Syrup Urine Disease

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

HEREDITY

- Autosomal Dominant Disorders
  - Can occur if only one person carries the gene for a particular disorder.
  - Affect males and females equally.
  - Results in physical/structural abnormalities.

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

HEREDITY

- Autosomal Dominant Disorders
  - Neurofibromatosis
  - Tuberous Sclerosis
  - Achondroplasia

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

HEREDITY

- Sex Linked / X Linked Disorders
  - Associated with genes on the X chromosome of a female.
  - The mother's X chromosome with the particular gene is passed on to a son.
  - The son is affected with a disorder from this gene. The disorder can be physical or intellectual

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

HEREDITY

- Sex Linked / X Linked Disorders
  - Muscular Dystrophy
  - Fragile X Syndrome
  - Hemophilia
  - Color Blindness

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

GROWTH BEFORE BIRTH

- A variety of factors can affect normal development and impact the fetus resulting in physical or intellectual impairment /disability.
  - Malnutrition
    - Early pregnancy – hydrocephalus, spina bifida, or prematurity.
    - Late pregnancy – low birth weight

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

GROWTH BEFORE BIRTH

- Malformation / Physical Disability – may occur in 3% of all births
  - Genetic – 25%
  - Environmental – 10%
  - Unknown causes – 65%

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

GROWTH BEFORE BIRTH

- Teratogens – Any agent that can cause a disability or defect in development.
  - Effect is a result of the degree of severity and timing of the teratogens presence.
  - Abnormalities may appear as early as the stage of the fertilized egg.

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

GROWTH BEFORE BIRTH

- Environmental toxins in the first days of pregnancy have an all or nothing effect.
- The magnitude of the malformation is also affected by the amount of the teratogen encountered.
- Effects late in pregnancy are not on organ development by rather on the size and weight of the infant.

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

GROWTH BEFORE BIRTH

- Types of Teratogens
  - Radiation
    - Growth retardation – 2nd & 3rd months
    - Mental retardation – 4th & 5th months
    - Microcephaly – 4th & 5th months
    - Eye malformations – 4th & 5th months
    - Genital / Skeletal Malformations
    - Death – 1st, 2nd & 3rd months

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

GROWTH BEFORE BIRTH

- Types of Teratogens
  - Medications
    - Anticonvulsants (all seizure meds)
      - Craniofacial Defects
      - Mental Retardation
      - Growth Retardation
      - Cardiovascular Defects
      - Spina Bifida

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

GROWTH BEFORE BIRTH

- Medications (cont'd)
  - Anticancer drugs
    - Hydrocephalus
    - Digital Abnormalities
    - Growth Retardation
    - Cardiovascular Defects
    - Spina Bifida
    - Miscarriage

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

GROWTH BEFORE BIRTH

- Medications (cont'd)
  - Anti-clotting medications
    - Digital Abnormalities
    - Mental Retardation
    - Low Birth Weight
  - Acne Medication (Acutane)
    - Brain malformations
    - Craniofacial Malformations

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

GROWTH BEFORE BIRTH

- Infections
  - STORCH Infections
    - Syphilis
    - Toxoplasmosis
    - Rubella
    - Cytomegalovirus (CHMV)
    - Herpes Virus

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

GROWTH BEFORE BIRTH

- Infections (cont'd)
  - Effects of Infections
    - Mental Retardation-T,C,R
    - Neurological Deficits-C
    - Micro/hydrocephalus-T,C,R,H
    - Hearing/Vision Loss-T,C,R
    - Cardiovascular Defects-S,R

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

PRENATAL DIAGNOSIS

- Hundreds of genetic disorders are diagnosable in utero.
- Reasons for prenatal testing:
  - Advanced maternal age.
    - 20-25 yrs. – 1 in 2,000
    - 35 yrs. – 1 in 400
    - 45 yrs – 1 in 32
  - Previous pregnancy with disability.

CONDITIONS RESULTING IN PHYSICAL DISABILITIES

PRENATAL DIAGNOSIS

- Methods of prenatal testing.
  - Amniocentesis
  - Chronic Villus Sampling
  - Alpha-fetoprotein Testing
  - Ultrasonography
  - DNA Analysis
  - DNA Analysis continued

Discussion Question 1

Conditions Resulting in Physical Disabilities

Given our current knowledge base regarding genetic transmission of disabilities and medical science's ability to diagnose disability during prenatal periods is it ethical and or moral for parents to terminate a pregnancy simply for the purposes of not having a child with a disability?

Quiz 1
Conditions Resulting in Physical Disabilities

1. Which of the following is not a characteristic of autosomal dominant disorders?

   A. Affects males and females equally
   B. Both parents must carry the gene
   C. Risk is low in affected families
   D. Results in physical / structural anomalies

2. Which of the following is not a characteristic of autosomal recessive disorders?

   A. Affects males and females equally
   B. Only one parent must carry the gene
   C. Risk is high in affected families
   D. Results in biochemical/enzymatic anomalies

3. Which of the following means of prenatal testing involves the withdrawal of a small amount of fluid for testing?

   A. Chorionic villus sampling
   B. Maternal serum alfa-fetoprotein testing
   C. Amniocentesis
   D. Ultrasonography

4. Which of the following means of prenatal testing involves the sampling of a small piece of lining from the placenta?

   A. Chorionic villus sampling
   B. Maternal serum alfa-fetoprotein testing
   C. Amniocentesis
   D. Ultrasonography

5. Which of the following means of prenatal testing involves the withdrawal of a small amount of blood to test?

   A. Chorionic villus sampling
   B. Maternal serum alfa-fetoprotein testing
   C. Amniocentesis
   D. Ultrasonography

6. Which of the following means of prenatal testing involves the use of sound waves to create a picture of the fetus?

   A. Chorionic villus sampling
   B. Maternal serum alfa-fetoprotein testing
   C. Amniocentesis
   D. Ultrasonography

7. X linked disorders are associated with genes on which of the following chromosome?

   A. X female chromosome
   B. X male chromosome
   C. Y female chromosome
   D. Y male chromosome

8. Which of the following is not a STORCH infection?

   A. Rubella
   B. Cytomegalovirus
   C. Toxoplasmosis
   D. Rubiola

9. Which of the following occurs during cell division and results in both chromosomes going to one cell instead of the one to one split?

   A. Translocation
   B. Deletion
   C. Non-disjunction
   D. Dialation

10. Which of the following is problem occurring during cell division and results in a portion of a chromosome breaking off?

   A. Translocation
   B. Deletion
   C. Non-disjunction
   D. Dialation