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Title:

MENDELS'S LAW OF SEGREGATION VALIDATED THROUGH PREIMPLANTATION GENETICS DIAGNOSIS

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Background:

Between 1865 and 1866, Gregor Mendel conducted hybridization experiments in garden peas from which he induced two genetic generalizations (Principle of Segregation and Independent Assortment) that later became Mendel's Principles of Heredity.

Objective:

With the ability to now identify embryonic allelic expression using preimplantation genetic diagnosis (PGD), this study analyzed whether the theoretical Law of Independent Assortment can be corroborated by actual genomic results.

Materials and Methods:

The retrospective study included all patients undergoing IVF with PGD for single gene disorder (SGD) with a recessive inheritance pattern carried out from January 1, 2010 to November 30, 2015 were included in the study. Genetic results were interpreted as Affected, Unaffected or Carrier. The proportion of each group was documented.

Result(s):

Eighty one patients underwent IVF/PGD for SGD; from which 569 embryos were tested for a 19 different diseases (Table 1). Carrier (47.3% (n=279)), Affected (28.1% (n=160)) and Unaffected (24.6% (n=125)) allelic reports were identified.

Conclusion(s):
According to Mendel's Laws, each screened embryo has a 25% chance of inheriting two abnormal genes, a 25% chance of inheriting two normal genes, and a 50% chance of inheriting a normal and abnormal gene. After analyzing over 500 embryos, this study validates Mendel's Laws of Segregation, a remarkable testament to a concept theorized over 150 years ago.

Support:
None

References:
None

Table 1 :

Disease	N-value	Percentage
Cystic Fibrosis	232	0.408
Gaucher Disease	71	0.125
Spinal Muscular Atrophy	38	0.067
Fanconi anemia, complementation group C	36	0.063
Familial Dysautonomia	35	0.062
Myotonia Congenita	30	0.053
Nonsyndromic Hearing Loss	29	0.051
Polycystic Kidney Disease	21	0.037
Senior-Loken Syndrome	17	0.030
Glycogen Storage Disease 1a	11	0.019
Smith Lemli Optiz Syndrome	11	0.019
Osteogenesis Imperfecta Type 1	8	0.014
Sickle Cell Anemia	8	0.014
Mitochondrial DNA depletion Syndrome 7	7	0.012
Congenital Adrenal Hyperplasia	5	0.009
Familial Mediterranean Fever	5	0.009
Usher Syndrome Type III	3	0.005
Congenital Disorder of Glycosylation	1	0.002
Mucolipidosis Type IV	1	0.002
Total	569	1