CONTENTS

	Page
List of tables	II
List of figures	IV
Introduction	1
Aim of work	2
Review of literature	3
Material and methods	30
Results	35
Discussion	133
Summary and conclusion	163
Recommendation	165
References	167
Arabic summary	186

List of Tables

No		page
1	Phenotypic Features of Trisomy 21 Syndrome (Down syndrome)	19
2	Phenotypic Feature of Trisomy 18 Syndrome (Edward syndrome)	22
3	Phenotypic Features of Trisomy 13 Syndrome (Patu syndrome)	24
4	Cases count of Normal and Abnormal karyotype patients with the sex	43
	difference of each type.	
5	Normal karyotype cases count (46,XX & 46,XY) summaries of sex type, consanguinity type, age of mother, age of father and order in family.	43
6	Abnormal karyotype cases count summaries of sex type, consanguinity	46
U	type, age of mother, age of father and order in family.	40
7	All karyotype cases count summaries of sex type, consanguinity type, age of mother, age of father and order in family.	49
8	Cases count of Abnormal karyotype patients with the sex difference of each type	52
9	Down's syndrome cases count summaries of sex type, consanguinity type, age of mother, age of father and order in family	55
10	Correlation between age of mother and incidence of their Trisomy 21 offspring's.	55
11	Correlation between age of father and incidence of their Trisomy 21 offspring's.	55
12	Edward's syndrome cases count summaries of sex type, consanguinity type, age of mother, age of father and order in family.	62
13	Patu's syndrome cases count summaries of sex type, consanguinity type, age of mother, age of father and order in family.	68
14	Turner's syndrome cases count summaries of sex type, consanguinity type, age of mother, age of father and order in family.	73
15	Correlation between age of mother and incidence of their Turner's offspring's.	73
16	Correlation between age of father and incidence of their Turner's offspring's.	73
17	Klinefelter's syndrome cases count summaries of sex type, consanguinity	79
	type, age of mother, age of father and order in family.	
18	Other's chromosomal aberrations cases count summaries of sex type, consanguinity type, age of mother, age of father and order in family.	83
19	Cases count of Normal karyotype patients with the sex difference of each	90
	type.	
20	Short stature cases count summaries of sex type, consanguinity type, age	93
	of mother, age of father and order in family.	

0.1		0.2
21	Correlation between age of mother and incidence of their Short stature	93
	offspring's.	0.2
22	Correlation between age of father and incidence of their Short stature	93
22	offspring's.	
23	Ambiguous genetalia cases count summaries of sex type, consanguinity	99
2.4	type, age of mother, age of father and order in family.	
24	Correlation between age of mother and incidence of their Ambiguous	99
2.7	genetalia offspring's.	
25	Correlation between age of father and incidence of their Ambiguous	99
26	genetalia offspring's.	404
26	The different normal cases count summaries of sex type, consanguinity	104
	type, age of mother, age of father and order in family.	404
27	Correlation between age of mother and incidence of the different normal	104
40	offspring's.	104
28	Correlation between age of father and incidence of the different normal	104
20	offspring's.	100
29	Comparison between Normal and Abnormal cases thorough the mother's	109
20	age	100
30	Comparison between Normal and Abnormal cases thorough the father's	109
21	age	111
31	Comparison between Normal and Abnormal cases thorough the	111
22	consanguinity and non-consanguinity.	110
32	Distribution of consanguinity of all karyotype cases.	113
33	Comparison between Normal and Abnormal cases thorough the sex type.	115
34	Comparison between Normal and Abnormal cases thorough the Order in	117
25	family.	110
35	Autosomal trisomic cases count (Down, Edward and Patu	119
	syndromes)summaries of sex type, consanguinity type, age of mother, age	
26	of father and order in family. Non trigomic coses count (Abnormal phonetyme and/or construe)	122
36	Non-trisomic cases count (Abnormal phenotype and/or genotype)	122
	summaries of sex type, consanguinity type, age of mother, age of father	
27	and order in family. Comparison between Trisomic and Non trisomic cases thorough the	125
37	Comparison between Trisomic and Non-trisomic cases thorough the mother's age.	125
38	Comparison between Trisomic and Non-trisomic cases thorough the	125
30	father's age.	143
39		127
37	Comparison between Trisomic and Non-trisomic cases thorough the	14/
40	consanguinity and non-consanguinity. Comparison between Trisomic and Non trisomic cases thorough the say.	129
40	Comparison between Trisomic and Non-trisomic cases thorough the sex	149
11	type.	101
41	Comparison between Trisomic and Non-trisomic cases thorough the	131
	Order in family.	

List of Figures

No		Page
1	The consanguinity type of Normal karyotype cases	44
2	The sex types of Normal karyotype cases	44
3	The order in family of Normal karyotype cases	44
4	Mother's age of Normal karyotype cases	45
5	Father's age of Normal karyotype cases	45
6	Comparison between mother's and father's ages of their Normal	45
	karyotype offspring's	
7	The consanguinity type of Abnormal karyotype cases	47
8	The sex types of Abnormal karyotype cases	47
9	The order in family of Abnormal karyotype cases	47
10	Mother's age of Abnormal karyotype cases	48
11	Father's age of Abnormal karyotype cases	48
12	Comparison between mother's and father's ages of their Abnormal	48
	karyotype offspring's	
13	The consanguinity type of All karyotype cases	50
14	The sex types of All karyotype cases	50
15	The order in family of All karyotype cases	50
16	Mother's age of All karyotype cases	51
17	Father's age of All karyotype cases	51
18	Comparison between mother's and father's ages of All karyotype	51
	offspring's	
19	Abnormal karyotype cases count	53
20	Abnormal karyotype cases count through comparison between sex	53
	difference of each one	
21	A case of Down's syndrome patients.	54
22	The metaphase spreads (A) and its karyotype (B) of non-disjunction type	56
	of female Down's syndrome (47,XX,+21)	
23	The metaphase spreads (A) and its karyotype (B) of non-disjunction type	57
	of male Down's syndrome (47,XY,+21)	
24	The metaphase spreads (A) and its karyotype (B) of translocation type of	58
	female Down's syndrome(46,XX,t(14;21)+21)	
25	The consanguinity type of Down's syndrome cases	59
26	The sex types of Down's syndrome cases	59
27	The order in family of Down's syndrome cases	59
28	Mother's age of Down's syndrome cases	60
29	Father's age of Down's syndrome cases	60
30	Comparison between mother's and father's ages of their Down's	60
	syndrome offspring's	

31	A case of Edward's syndrome patients.	61
32	The metaphase spreads (A) and its karyotype (B) of non-disjunction type	63
	of female Edward's syndrome (47,XX, +18)	00
33	The metaphase spreads (A) and its karyotype (B) of non-disjunction type	64
	of male Edward's syndrome (47,XY, +18)	
34	The consanguinity type of Edward's syndrome cases	65
35	The sex types of Edward's syndrome cases	65
36	The order in family of Edward's syndrome cases	65
37	Mother's age of Edward's syndrome cases	66
38	Father's age of Edward's syndrome cases	66
39	Comparison between mother's and father's ages of their Edward's	66
	syndrome offspring's	
40	A case of Patu's syndrome patient.	67
41	The metaphase spreads (A) and its karyotype (B) of non-disjunction type	69
	of female Patu's syndrome (47, XX, +13)	
42	The consanguinity type of Patu's syndrome cases	70
43	The sex types of Patu's syndrome cases	70
44	The order in family of Patu's syndrome cases	70
45	Mother's age of Patu's syndrome cases	71
46	Father's age of Patu's syndrome cases	71
47	Comparison between mother's and father's ages of their Patu's syndrome	71
	offspring's	
48	A case of Turner's syndrome patients.	72
49	The metaphase spreads (A) and its karyotype (B) of isochromosome X	74
	type of Turner's syndrome (46,Xi(X)(q10))	
50	The metaphase spreads (A) and its karyotype (B) of monosomy X type	75
	of Turner's syndrome (45,XO)	
51	The consanguinity type of Turner's syndrome cases	76
52	The sex types of Turner's syndrome cases	76
53	The order in family of Turner's syndrome cases	76
54	Mother's age of Turner's syndrome cases	77
55	Father's age of Turner's syndrome cases	77
56	Comparison between mother's and father's ages of their Turner's	77
	syndrome offspring's.	
57	A case of Klinefelter's syndrome patients.	78
58	The metaphase spreads (A) and its karyotype (B) of Klinefelter's	80
	syndrome (47,XXY)	
59	The consanguinity type of Klinefelter's syndrome cases	81
60	The sex types of Klinefelter's syndrome cases	81
61	The order in family of Klinefelter's syndrome cases	81
62	Mother's age of Klinefelter's syndrome cases	82

63	Father's age of Klinefelter's syndrome cases	82
64	Comparison between mother's and father's ages of their Klinefelter's	82
V-7	syndrome offspring's	02
65	The metaphase spreads (A) and its karyotype (B) of (47,XXX) case	84
66	The metaphase spreads (A) and its karyotype (B) of (48,XXY,+21) case	85
67	The metaphase spreads (A) and its karyotype (B) of (46,XY,inv(5)(p))	86
	case	
68	The metaphase spreads (A) and its karyotype (B) of (46,X,inv(Y)) case	87
69	The consanguinity type of Other's chromosomal aberrations cases	88
70	The sex types of Other's chromosomal aberrations cases	88
71	The order in family of Other's chromosomal aberrations cases	88
72	Mother's age of Other's chromosomal aberrations cases	89
73	Father's age of Other's chromosomal aberrations cases	89
74	Comparison between mother's and father's ages of their Other's chromosomal aberrations offspring's.	89
75	Comparison between each type of Normal (Normal, Short stature, and Ambiguous genetalia) and Abnormal (Down, Edward, Patu, Klinefelter, Turner, and other's chromosomal aberrations) karyotype cases.	91
76	A case of Short stature syndrome patient.	92
77	The metaphase spreads (A) and its karyotype (B) of female short stature case (46,XX)	94
78	The metaphase spreads (A) and its karyotype (B) of male short stature case (46,XY)	95
79	The consanguinity type of Short stature cases	96
80	The sex types of Short stature cases	96
81	The order in family of Short stature cases	96
82	Mother's age of Short stature cases	97
83	Father's age of Short stature cases	97
84	Comparison between mother's and father's ages of their Short stature offspring's.	97
85	A case of Ambiguous genetalia syndrome patients.	98
86	The metaphase spreads (A) and its karyotype (B) of female Ambiguous	100
07	genetalia case (46,XX)	101
87	The metaphase spreads (A) and its karyotype (B) of male Ambiguous genetalia case (46,XY)	101
88	The consanguinity type of Ambiguous genetalia cases	102
89	The sex types of Ambiguous genetalia patient	102
90	The order in family of Ambiguous genetalia cases	102
91	Mother's age of Ambiguous genetalia cases	103
92	Father's age of Ambiguous genetalia cases	103
93	Comparison between mother's and father's ages of their Ambiguous	103

	genetalia offspring's.	
94	The metaphase spreads (A) and its karyotype (B) of female different	105
	normal case (46,XX)	100
95	The metaphase spreads (A) and its karyotype (B) of male different	106
	normal case (46,XY)	
96	The consanguinity type of different normal cases	107
97	The sex types of different normal cases	107
98	The order in family of different normal cases	107
99	Mother's age of different normal cases	108
100	Father's age of different normal cases	108
101	Comparison between mother's and father's ages of the different normal	108
	offspring's.	
102	Comparison between Normal and Abnormal karyotype cases thorough	110
	the mother's age.	
103	Comparison between Normal and Abnormal karyotype cases thorough	110
	the father's age.	
104	Comparison between Consanguinity and Non-consanguinity of normal	112
	and abnormal karyotype cases.	
105	Comparison between Normal and Abnormal karyotype cases through	112
	consanguinity and non-consanguinity.	
106	Comparison between Consanguinity and Non-consanguinity of normal	112
10=	and abnormal karyotype cases	
107	The distribution of Consanguinity and Non-consanguinity of all	114
100	karyotype cases	117
108	Comparison between Normal and Abnormal karyotype cases through	116
100	sex type	117
109	Comparison between Males and Females of normal and abnormal	116
110	karyotype cases Comparison between Normal and Abnormal karyotype cases through	110
110	Comparison between Normal and Abnormal karyotype cases through the order in family	118
111	The consanguinity type of Autosomal trisomic cases	120
112	The sex types of Autosomal trisomic cases	120
113	The order in family of Autosomal trisomic cases	120
114	Mother's age of Autosomal trisomic cases	121
115	Father's age of Autosomal trisomic cases	121
116	Comparison between mother's and father's ages of their Autosomal	121
	trisomic offspring's.	
117	The consanguinity type of Non-trisomic cases	123
118	The sex types of Non-trisomic cases	123
119	The order in family of Non-trisomic cases	123
120	Mother's age of Non-trisomic cases	124

121	Father's age of Non-trisomic cases	124
122	Comparison between mother's and father's ages of their Non-trisomic	124
	offspring's	
123	Comparison between Trisomic and Non-trisomic cases thorough the	126
	mother's age	
124	Comparison between Trisomic and Non-trisomic cases thorough the	126
	father's age	
125	Comparison between Trisomic and Non-trisomic through consanguinity	128
	and non-consanguinity	
126	Comparison between Consanguinity and Non-consanguinity of trisomic	128
	and non-trisomic cases	
127	Comparison between Trisomic and Non-trisomic cases through sex type	130
128	Comparison between Males and Females of trisomic and non-trisomic	130
	cases	
129	Comparison between Trisomic and Non-trisomic through the order in	132
	family.	