مستشفى الملك فيصل التخصصي ومركز الأبحاث
King Faisal Specialist Hospital & Research Centre

Rare Dental Disorders Registry "RDDR"

The 1st Accumulative Annual Report

2011 - 2022

Annual Report prepared by the staff of the Dental Department and Research Centre, Biostatistics Epidemiology & Scientific Computing Department

King Faisal Specialist Hospital and Research Centre

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Foreword

The information presented and analyzed in this report was gathered over three stages of meetings and investigations. The first phase was carried out between 2004 and 2006, prior to the establishment of the registry, with the collaboration of two Swedish centers (Gothenburg & Jonkoping). The second phase began in 2011 with the official approval of the Rare Dental Disorder Registry, and the third phase began in 2016 with electronic documentation and supplementing the first demographic data acquisition with the assistance of excellent dental and core registry teams.

I am grateful to KFSHRC and the dental team, to name a few: Drs. Hassa Hansson, Richard Hakansson, Christer Henningson, Khalid AlZoman, Moh'd AlHelal, and Dental Assistance Rhenee Torres. Using the five different approved documentation forms for examining, treating, chart reviewing, and data capturing over the years. Dr. Edward Devol, Chairman of Biostatistics, Epidemiology, and Scientific Computing, and Ms. Abeer AlFirm, Associate Research Data Manager, and Ms.Manal AlMarzouqi, Head of Disease Registries. For establishing and updating the registry in electronic format, cleaning the data from the Cerner MHR, and communicating with all team members over the years.

Dr. Adeeb Al Omrani

Principal investigator

Acknowledgments

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Papillion Lefèvre Syndrome

Papillon-Lefèvre syndrome (PLS): “Is an extremely rare genetic disorder that typically becomes apparent from approximately one to five years of age”.

It develops a dry scaly patches on the skin of the palms and the soles, and associated with severe inflammation and degradation of the structures surrounding the teeth.

PLS’s complications: frequent skin infections, abnormalities of the nails and excessive perspiration (hyperhidrosis).

Papillon-Lefèvre syndrome is inherited in an autosomal recessive pattern. It results from changes (alterations) of the CTSC gene that regulates production of an enzyme known as cathepsin C.

https://rarediseases.org/rare-diseases/papillon-lefevre-syndrome/

Ectodermal Dysplasiamal Dysplasia

Ectodermal dysplasias (ED) are “disorders that affect the skin, sweat glands, hair, teeth, and nails. Some individuals with ED may also have cleft lip and/or palate. ED can additionally cause problems with the immune system as well as hearing and vision. There are more than 180 specific types have been identified”.

As well as ED a rare disease, it affects fewer than 200,000 people in US.

Ectodermal dysplasias occur when the outer layer of tissue (ectoderm) of the embryo does not develop normally. If two or more body structures derived from the ectoderm are affected, a person is considered to have ED.

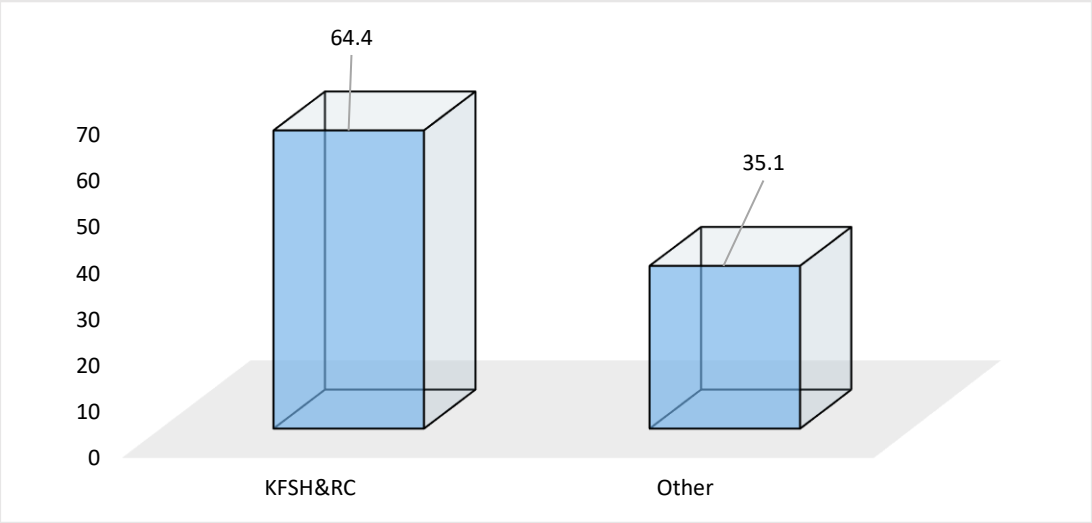
Ectodermal dysplasias are caused by a change, or mutation, in a gene. It can be hereditary or not.

Ectodermal Dysplasia | National Institute of Dental and Craniofacial Research (nih.gov) /

1.0 Demographic

1.1 Referring Center:

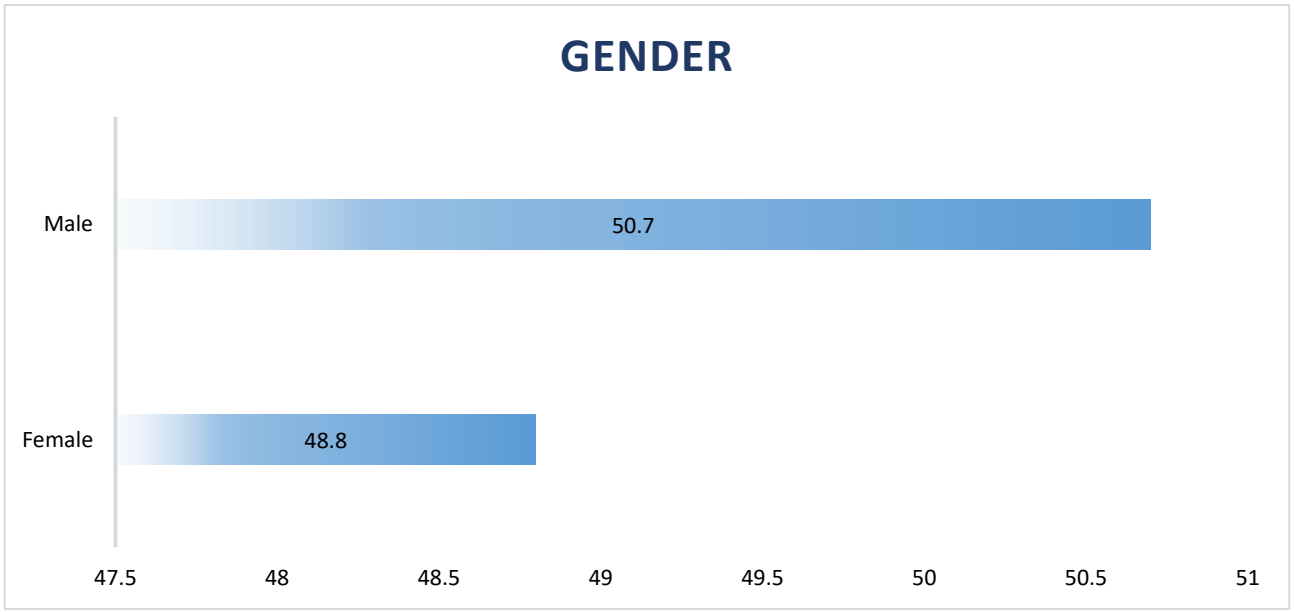
The total number of patients who were referred to the Dental Department within King Faisal Specialized Hospital and Research Center were 132 patients (64.7%) and a count of 72 patients (35.3%) were referred from Other Referring Centers.



Graph 1.1 Number of Referring Center

1.2 Gender:

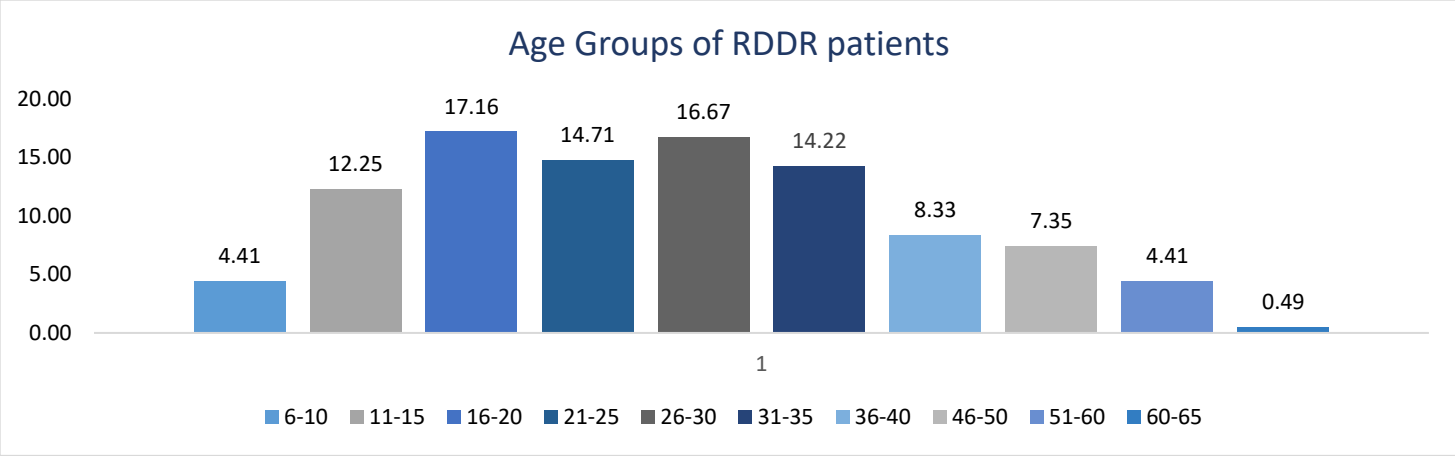
The total population presented to RDDR are 204 patients, with a count of 104 males (51%) and 100 females (49%).



Graph 1.2 Number of gender

1.3 Age Groups in RDD Patients:

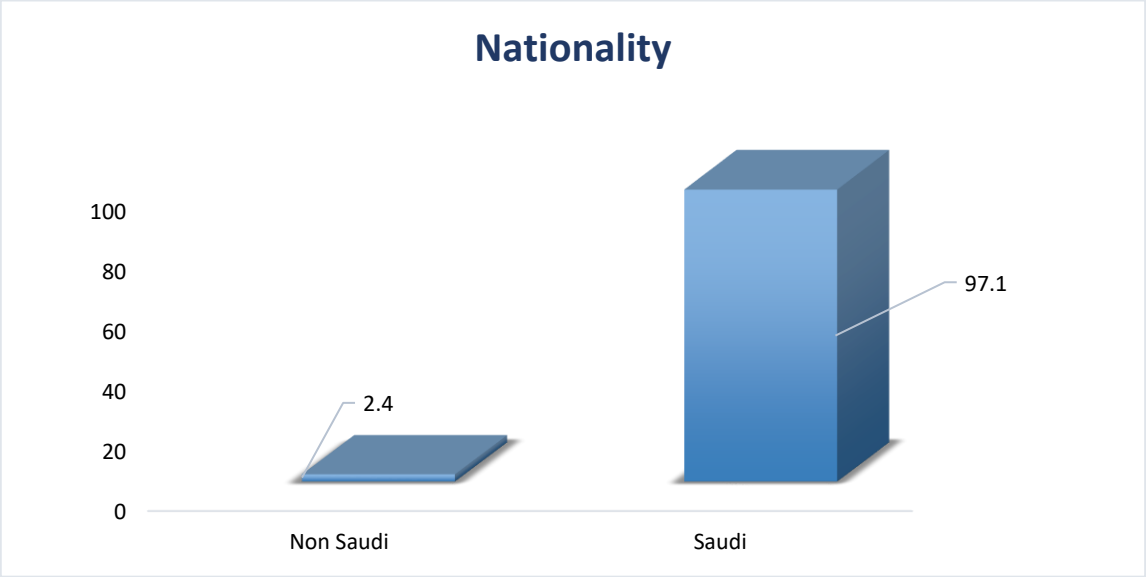
Among all 204 patients registered in the RDD, the highest age group are allocated in 16-20 group with a count of 35 patients (17.16%). Age group 26-30 came in second with a count of 34 patients (16.67%). Age group 60-65 came in last with a number of one patient (0.49%).



Graph 1.3 Number of Age Groups in RDD Patients

1.4 Nationality

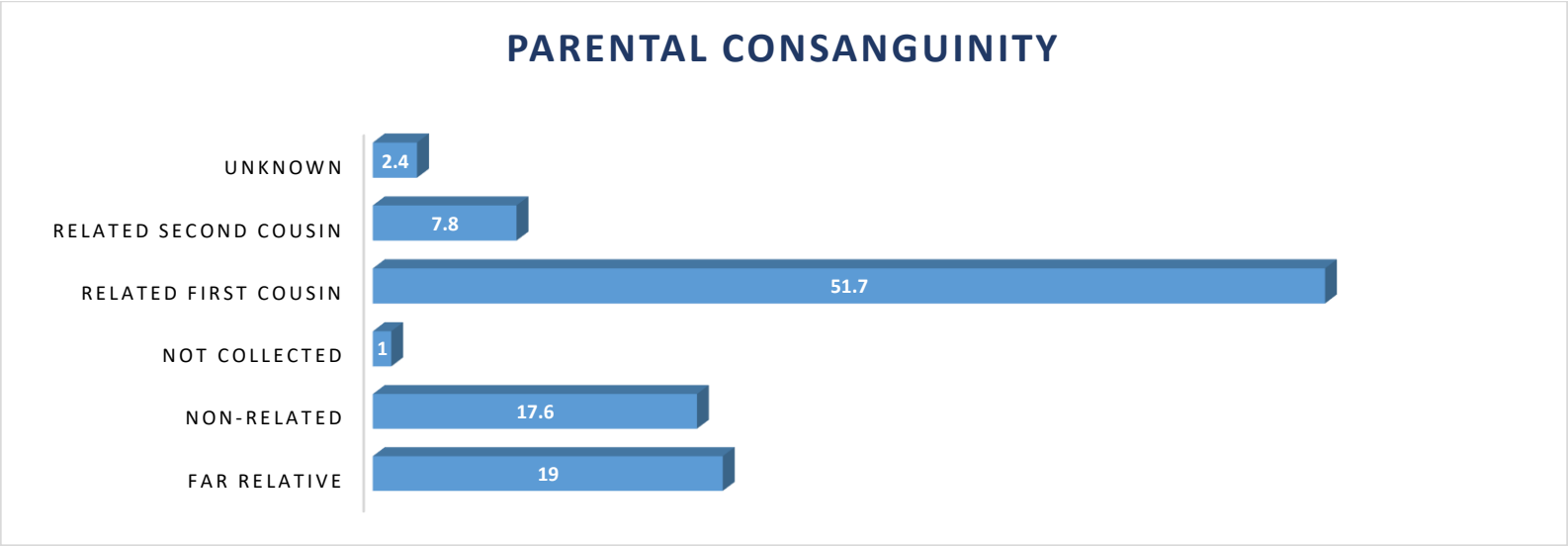
The patient population of the RDD reported. With a count of 204 patients. Majority of the patients presented were Saudi, with a number of 199 (97.5%), and a number of 5 (2.5%) were non-Saudi.



Graph 1.4 Number of Nationality

1.5 Parental Consanguinity

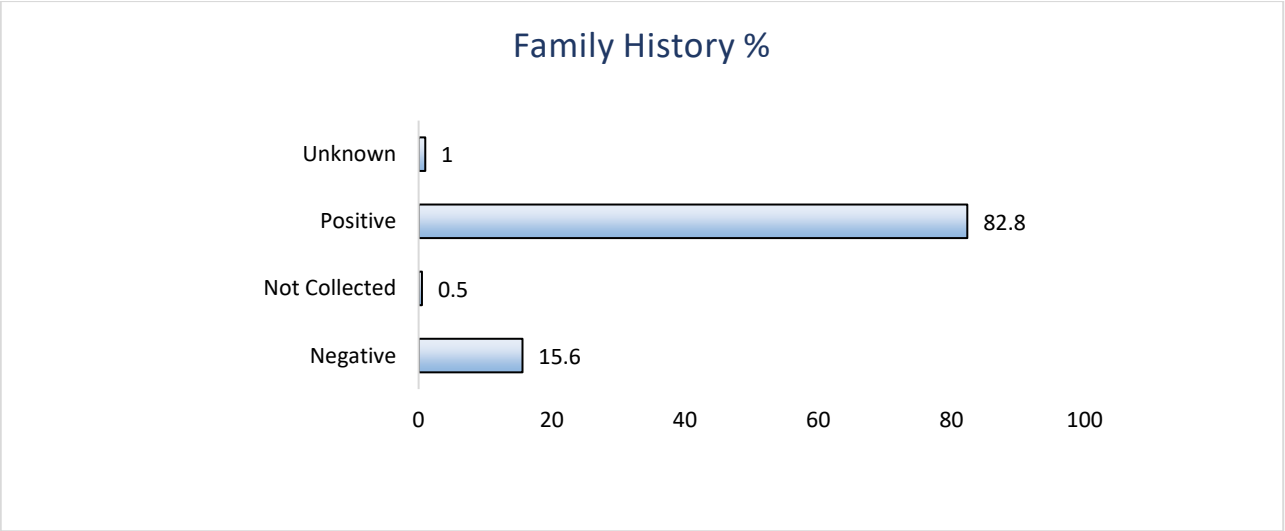
Demonstrated in the demographic of the RDDR, more than half of the number of patients registered in the First Cousin Relation with a count of 106 (52%), As per the remaining counts, a number of 7 patients either refused to report or they were out of reach (3.5%) for eligibility issues or lost of follow ups.



Graph 1.5 Number of Parental Consanguinity

1.6 Family History

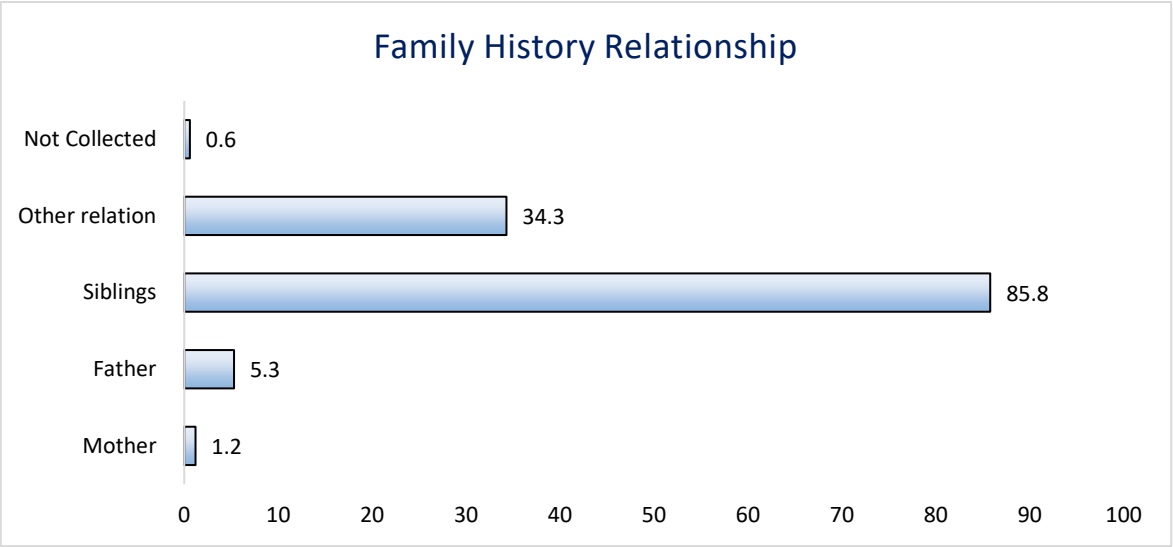
Demonstrated in the RDDR with a total count of 204 patients, most patients reported positive with a count of 169 (82.8%), and quarter stated negative with a number of 32 patients (15.6%). Moreover, 3 patients either refused to report or they were out of reach (2.5%).



Graph 1.6 Number of Parental Consanguinity

1.7 Family History Relationship

It is fair to say that most of the family History Relationship is shown in Siblings in 145 (85.8%). The Other relations such as cousins, uncles, and aunties are common to be reported with a count of 58 (34.4%).



Graph 1.7 Number of Family History Relationship

1.8 Number of Siblings

One sibling being affected in each patient has a count of 65 (44.8%). At least four siblings being affected in each patient has a count of 11 (7.6%). As per five siblings being affected in each patient has, a count of zero has being reported.

Number of Siblings	Frequency	Percent
With One effect Sibling	65	31.7
With Two effect Siblings	43	21
With Three Two effect Siblings	26	12.7
With Four Two effect Siblings	11	5.4
Total	145	70.7

Table 1.1 Number of Siblings

1.9 Number of Other Relation

The Other relations as grandfathers, cousins, uncles, offerings, and aunties are common in any family history. A total number of 59 (28.92%) in the over batch of other relations. One relation has been affected with a count of 25 (12.2%). In the Five relations, a count of 6 (2.9%) has been reported.

Number of Other Relation	Frequency	Percent
With One effected Other Relation	25	12.2
With Two effected Other Relation	11	5.4
With Three effected Other Relation	5	2.4
With Four effected Other Relation	11	5.4
With Five effected Other Relation	6	2.9
Total	**58**	28.3
With No effected Other Relation	147	71.7
Total	**205**	100

Table 1.2 Number of Other Relation

1.10 Place of Birth

Demonstrated in the demographic of the RDDR out of 204 patients, a number of 121 (59.31%) were born in Riyadh Region. Whereas a count of 1 (0.49) and 2 patients with (0.98 %) were all born outside the Kingdom of Saudi Arabia with minimal numbers.

Country	City	N	%
GCC	Kuwait	6	2.94
SA	Riyadh Region	121	59.31
SA	Makkah Region	15	7.35
SA	Al Madinah Region	10	4.9
SA	Al-Qassim Region	3	1.47
SA	Eastern Province	29	14.22
SA	'Asir Region	4	1.96
SA	Tabuk Region	1	0.49
SA	Jizan Region	3	1.47
SA	Najran Region	8	3.92
SA	Al Bahah Region	1	0.49
Arab County	Lebanon	1	0.49
European Country	Australia	2	0.98
	Total	**204**	100.00

Table 1.3 Number of Patients Place of Birth

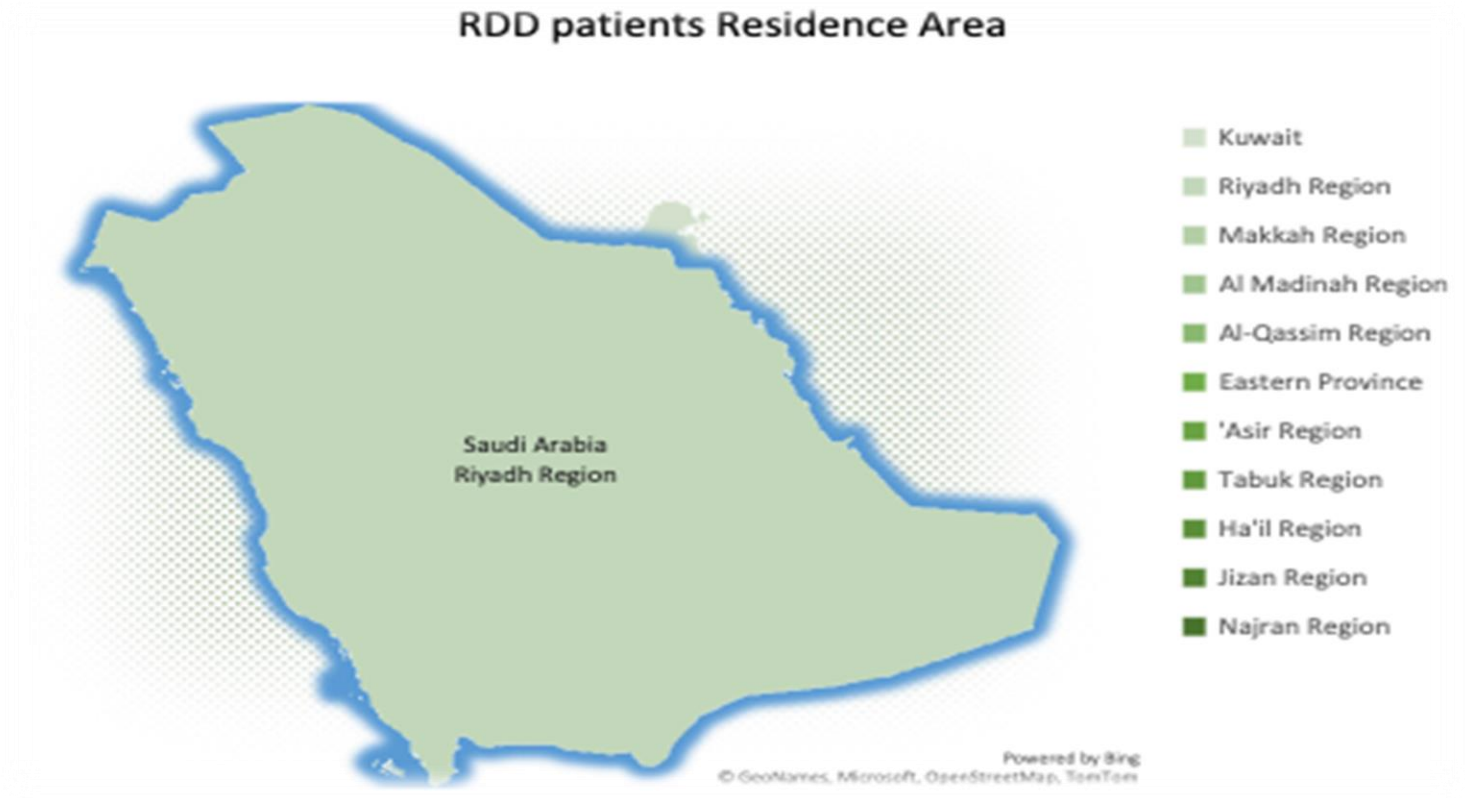
Graph 1.8 Number of Patients National Place of Birth

1.11 Residence Area

Demonstrates the demographic of the RDDR in total of patients 204, Riyadh Region had the highest ranking of all regions with a number of 124 (60.78%). Kuwait has a number of 6 patients with (2.94%) of patients came from outside the Kingdom of Saudi Arabia.

Country	City	%	N
SA	Riyadh Region	60.78	124
SA	Eastern Province	13.73	28
SA	Makkah Region	6.37	13
SA	Al Madinah Region	4.9	10
SA	Najran Region	3.92	8
GCC	Kuwait	2.94	6
SA	'Asir Region	2.94	6
SA	Al-Qassim Region	1.4	3
SA	Jizan Region	1.47	3
SA	Ha'il Region	0.98	2
SA	Tabuk Region	0.49	1
	Total	100	**204**

Table 1.4 Number of Patients Residence Area



Graph 1.9 Distribution of Patients Residence Area

2.0 Clinical Data

2.1 RDD Over all reported Diagnosis

Table displayed shows the number times diseases were selected for the 204 patients with a total number of 209. This is an indication that one or more RDD patient has been diagnosed with more than one illness.

Over all Diagnosis	N	%
Papillon Lefevre Syndrome	136	66.7
Ectodermal Dysplasia	58	28.4
Cleidocranial Dysplasia	3	1.5
Others	12	5.9
Total	**209**	102.5

Table 1.5 Number of RDD Over all reported Diagnosis

2.2 Diagnosis (choice=Papillon Lefevre Syndrome)

Table displayed shows the number of patients who were diagnosed with Papillon Lefevre Syndrome, with a count of 136 (66.7%) and a number of 68 (33.3%) were not.

Papillon Lefevre Syndrome	Frequency	Percent
Yes	136	66.7
No	68	33.3

Table 1.6 Number of Patients with Papillon Lefevre Syndrome

2.3 Diagnosis (choice=Ectodermal Dysplasia)

Table demonstrated in total of patients who were diagnosed with Ectodermal Dysplasia, showed a count of 146 (71.2%) of the patients were not diagnosed with Ectodermal Dysplasia, and a number of 58 (28.3%) of the collected patients were diagnosed.

Diagnosis (choice=Ectodermal Dysplasia)	Frequency	Percent
Yes	58	28.3
No	146	71.2

Table 1.7 Number of Patients with Ectodermal Dysplasia

2.4 Diagnosis (choice=Bone Diseases)

Table illustrate the Patients who diagnosed with Bone disease, (99.5%) almost all of the sample were not diagnosed with Bone disease, and (0.5%) of them were diagnosed.

Diagnosis (choice=Bone Diseases)	Frequency	Percent
Yes	1	0.5
No	204	99.5

Table 1.8 Number of Patients with Bone Diseases

2.5 Diagnosis (choice=Cleidocranial Dysplasia)

Table illustrated sowed patients who were not diagnosed with Cleidocranial Dysplasia. A very large count of 201 (98%). On the other hand, patients that were diagnosed showed a number of 3 (1.5%).

Diagnosis (choice=Cleidocranial Dysplasia)	Frequency	Percent
Yes	3	1.5
No	201	98

Table 1.9 Number of Patients with Cleidocranial Dysplasia

2.6 Diagnosis (choice=Anodontia)

Table display the Patients who diagnosed with Anodontia, (100%) almost all of the sample were not diagnosed with Anodontia, and zero of the sample were diagnosed.

Diagnosis (choice=Anodontia)	Frequency	Percent
Yes	0	0
No	204	100

Table 1.10 Number of Patients with Cleidocranial Dysplasia

2.7 Sub Diagnosis (choice=Others)

Table display the Patients who diagnosed with others sub diagnosis, (93.7%) most of the patients of this category were not diagnosed with others Sub diagnosis, while a count of 12 indicated with a (5.9%) to be true.

Diagnosis (choice=Others)	Frequency	Percent
Yes	12	5.9
No	192	93.7

Table 1.11 Number of Patients with who diagnosed with others sub diagnosis

2.8 Sub Diagnosis (choice=Amelogenesis Imperfecta)

Table illustrate shows almost all patients were not diagnosed with Amelogenesis Imperfecta. A number of 203 (99%) and 1 (0.5%) of them were diagnosed.

Diagnosis Sub (choice=Amelogenesis Imperfecta)	Frequency	Percent
Yes	1	0.5
No	203	99

Table 1.12 Number of Patients with Amelogenesis Imperfecta

2.9 Diagnosis Sub (choice=Deatinogenesis)

Table illustrate the patients who were diagnosed with Deatinogenesis. A count of 203 (99.5%) almost all of the patients were not diagnosed with Deatinogenesis. and 1 (0.5%) of the sample were diagnosed.

Diagnosis Sub (choice=Deatinogenesis)	Frequency	Percent
Yes	1	0.5
No	203	99.5

Table 1.13 Number of Patients with Deatinogenesis

2.10 Diagnosis Sub (choice=Osteoptrosis)

Table display the patients who were diagnosed with Osteoptrosis, a count of 204 (100%) almost all of the patients were not diagnosed with Osteoptrosis, and. No patients showed any presented affect in this category.

Diagnosis Sub (choice=Osteoptrosis)	Frequency	Percent
Yes	0	0
No	204	100

Table 1.14 Number of Patients with Osteoptrosis

2.11 Diagnosis Sub (choice=Pagets Disease)

Table display the patients who diagnosed with Pagets Disease, a count of 204 (100%) almost all of the patients were not diagnosed with Pagets Disease, and, No patients showed any presented affect in this category.

Diagnosis Sub (choice=Pagets Disease)	Frequency	Percent
Yes	0	0
No	204	100

Table 1.15 Number of Patients with Pagets Disease

2.12 Diagnosis Sub(choice=Hypophosphatasia)

Table illustrate the patients who diagnosed with Hypophosphatasia, account of 198 (96.6%), most of the patients were not diagnosed with Hypophosphatasia and 6 (2.9%) of them were affected.

Diagnosis Sub (choice=Hypophosphatasia)	Frequency	Percent
Yes	6	2.9
No	198	96.6

Table 1.16 Number of Patients with Hypophosphatasia

2.13 Diagnosis Sub (choice=Osteogenesis Imperfecta)

Table illustrates the patients who were diagnosed with Osteogenesis Imperfecta, with a count of 199 (97.6%) majority of them were not diagnosed to be affected. While a number of 5 (2.4%) were found to be affected.

Diagnosis Sub (choice=Osteogenesis Imperfecta)	Frequency	Percent
Yes	5	2.4
No	199	97.6

Table 1.17 Number of Patients with Osteogenesis Imperfecta

2.14 Diagnosis Sub (choice=Complete (Hypodontial))

Table displays the patients who diagnosed with Complete (Hypodontial), a count of 204 (99.5%), almost all of the patients were not diagnosed Complete (Hypodontial), and 1 (0.5%) of them were diagnosed.

Diagnosis Sub (choice=Complete (Hypodontial))	Frequency	Percent
Yes	1	0.5
No	203	99.5

Table 1.18 Number of Patients with Complete Hypodontial

2.15 Diagnosis Sub (choice=Partial (Hypodontial))

Table display the Patients who diagnosed with Partial (Hypodontial), (99.5%) almost all of the sample were not diagnosed with Partial (Hypodontial), and zero of the sample were diagnosed.

Diagnosis Sub (choice=Partial (Hypodontial))	Frequency	Percent
Yes	0	0
No	204	100

Table 1.19 Number of Patients with Partial Hypodontial

2.16 Other Anomalies

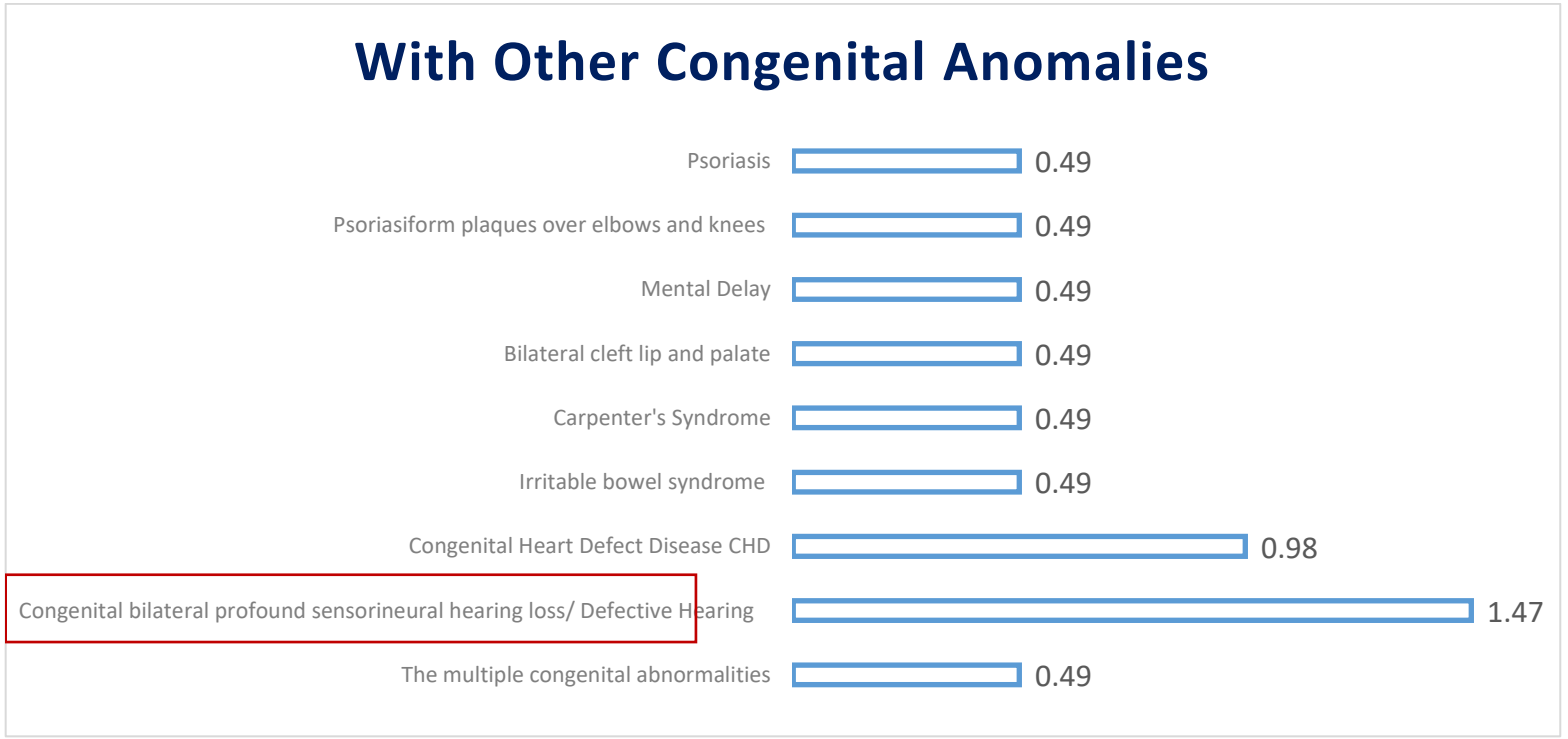
Table displays the patients who had other Anomalies that are Isolated, a count of 193 (99.6%), almost all of the patients reported to be true. While and 11 (5.4%) of them were with other Congenital Anomalies.

Other Anomalies	Frequency	Percent
Isolated	193	94.6
With other congenital Anomalies	11	5.4

Table 1.20 Number of Patients with other Anomalies

2.17 With Other Congenital Anomalies

As displays the patients who had other Anomalies with a count of 11 (5.4%) of them were with other Congenital Anomalies. The highest reported of them all is Congenital bilateral profound sensorineural hearing loss/Defective Hearing with a count of 3 (1.47%) of all collected in the RDD Registry.

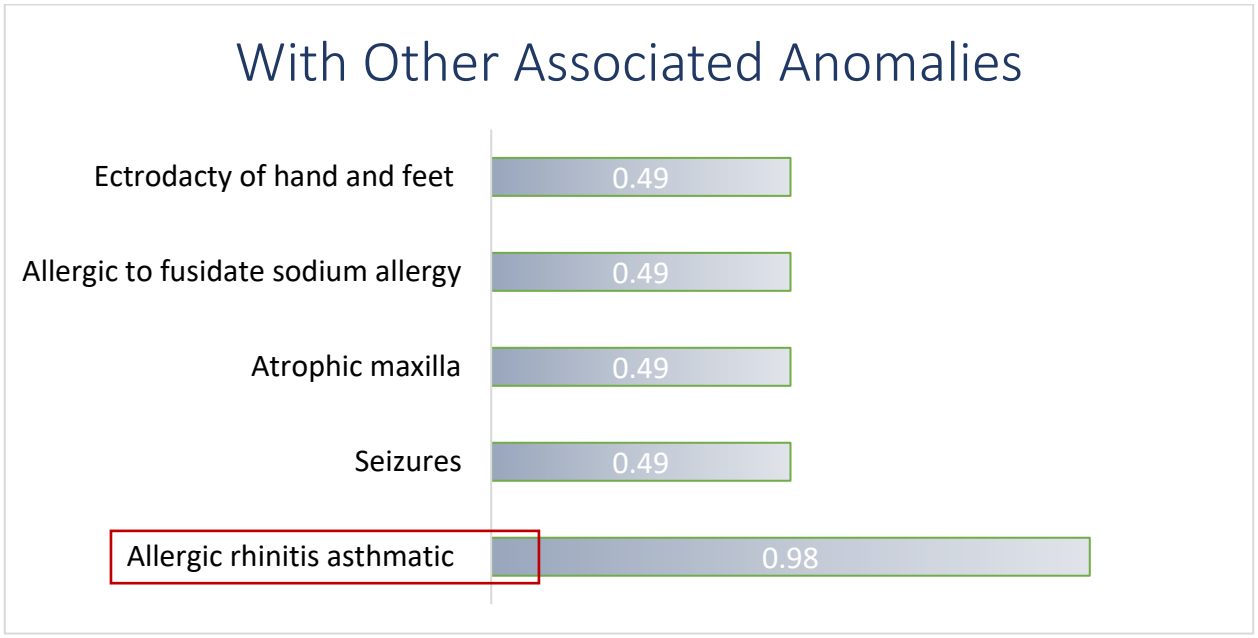


Graph 1.10 Distribution of Other Congenital Anomalies

2.18 With Other Associated Anomalies

As displays the patients who had other Anomalies with a count of 11 (5.4%) of them were with other Associated Anomalies.

The highest reported of them all is Asthma with a count of 2 (0.98%) of all collected in the RDD Registry.



Graph 1.11 Distribution of Other Associated Anomalies

A WORD TO THE WISE

"While alone we are here, but together we are powerful".

-Rare Disease Patients-


NORTH TOWER