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Original Article

# The clinical characteristics of Duane retraction syndrome in Al-Medina region

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## Abstract

*Purpose:* To assess the clinical characteristics of Duane Retraction Syndrome (DRS) in Al-Medina region, in patients attending a pediatric ophthalmology clinic at Ohud Hospital.
*Method:* A cross-sectional observational study was conducted from October 2017 to June 2018 at Ohud Hospital, Al-Medina region, Saudi Arabia. Data was collected using a sheet containing eighteen simple items which include demographic data, family history, surgical history, the clinical characteristics of the disease and the physician's treatment plan. Twenty patients were included in this study.
*Results:* All twenty patients included in the study were diagnosed with DRS type I. Male and female were symmetrical in numbers as both 10 (50.0%) respectively. The mean age of the subjects was 8.7 years (SD 9.6). 95% of the subject were unilaterally affected while five percent of them were affected both sides of the eyes. 75% of the patients were affected on the left eye while 20% were affected on the right side. 35% of the patients had amblyopia in the affected eye. 25% of the patients had an abnormal head position. Abnormal eye movement (upshoot and downshoot) more common when the right eye is affected ($p$-0.035). 75% of the patients were orthotropic while 30% had esotropia. 75% of patients were positive of parents' consanguinity. 16 patients (80.0%) were from Al-Medina city while four of them (20.0%) were from the different area of Al-Medina region (Al Hejeria, Al Henakia, Omluj, Yatmah).
*Conclusion:* This cross-sectional observational study is the first one that evaluates DRS in Al-Medina region. Associated abnormal eye movement more common when the right eye is affected. Parents' consanguinity may play a role in the occurrence of DRS as it is not considered as a rare disease in our region. Therefore, more studies are needed to establish the relationship between the occurrence of DRS and parents' consanguinity in our region. Also, further studies are needed to establish the relationship between abnormal eye movement and the involvement of the right eye.

**Keywords**: Duane retraction syndrome (DRS), Clinical characteristics, Saudi Arabia, Al-Medina


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## Introduction

Duane Retraction Syndrome (DRS) is a rare congenital paralytic strabismus. Although the etiology of DRS is unknown, three possible causes have been found. A mechanical, congenital neural and central nervous system abnormalities have been described.[1,2] According to Huber's classification, DRS has three different types: Type I described as a limited or

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complete absence of abduction with normal to slightly defective adduction; Type II reverse Type I, which described as a limited or complete absence of adduction with normal to near normal abduction; Type III is a combination of limitation to complete absence of both abduction and adduction.[1] DRS type I considered the most common type in both unilateral and bilateral cases followed by type II and III. In unilateral cases, left eye more frequently involved with female predominance, unlike bilateral DRS which is a male predominance.[3,4] Prevalence of amblyopia in DRS in unilateral cases is about 25–27% while in bilateral cases amblyopia is about 48%.[5,6] Recent studies have reported that α2-chimaerin (CHN1) missense mutations are the leading cause of DRS.[7,8] To the best of our knowledge, very few studies were done in Saudi Arabia about DRS.[3,4] These studies are old and there is no follow up information about the disease in the region. This is the first study from Al-Medina region that evaluates the DRS. The aim of this study is to evaluate the prevalence of DRS and its clinical characteristics in Al-Medina region of Saudi Arabia.

## Patients and Method

This study was approved by the research ethical approval committee at Ohud Hospital. A cross-sectional observational study was conducted from the period of October 2017 to June 2018 at Ohud Hospital in Al-Medina region. Clinical notes of the twenty patients presenting to the strabismus clinic during this period were studied. Twenty patients with DRS were identified and included in this study, ten patients were male and 10 were females (%50.0). The data was collected and reviewed by pediatric ophthalmologist. The information included demographic data, family history, surgical history, the clinical characteristics of the disease including laterality, Huber classification, amblyopia, abnormal head position, deviation in primary gaze, globe retraction, upshoot and downshoot. Other ocular congenital abnormalities, systemic congenital malformation, and treatment plan were also evaluated.

Statistical analysis: The data was collected on in excel and then was transported to statistical software SPSS Ver. 22 where both descriptive and inferential statistics were conducted. P-value of $\leq 0.05$ was accepted as the significance level for all statistical tests. All categorical variables presented in Tables 1 and 2 were summarized as counts and proportions (%) and mean ± standard deviation for all continuous variables. For categorical variable Chi-square test was used, and for continuous variables as independent t-test, was applied.

## Results

All patients included in our study were diagnosed with DRS type I. 20 patients with DRS were identified and included in the study, ten patients were males and 10 patients were females (50.0%). The mean age was 8.7 years (SD 09.6), the youngest age was 6 months while the oldest age was 28 years old. Table 1 presented the demographic characteristics of the patients. Ninety-Five percent of the subject were unilaterally affected while five percent had both eyes affected. 75% of the patients were affected on the left eye while 20% were affected on the right side. Table 2 summarized the clinical characteristics of the disease. 35% of the

**Table 1**. Socio demographic characteristics of patient (n=20).

| Study variables | N (%) |
|---|---|
| Age in years (mean ± standard deviation) | 08.7 ± 09.6 |
| *Gender* | |
| • Male | 10 (50.0%) |
| • Female | 10 (50.0%) |
| *Residency* | |
| • Al Madinah | 16 (80.0%) |
| • Outside Al Madinah | 04 (20.0%) |
| *Parents Consanguinity* | |
| • Positive | 14 (70.0%) |
| • Negative | 06 (30.0%) |
| *Positive Family History of Strabismus* | |
| • Yes | 06 (30.0%) |
| • No | 14 (70.0%) |
| *Onset of the disease* | |
| • Since birth | 15 (75.0%) |
| • 1 month–6 months | 03 (15.0%) |
| • >6 months–1 year | 02 (10.0%) |

**Table 2**. Clinical Characteristics of patient (n=20).

| Factor | N (%) |
|---|---|
| *Which eye is affected* | |
| • Right | 04 (20.0%) |
| • Left | 15 (75.0%) |
| • Both | 01 (05.0%) |
| Huber classification of left eye if affected – Type I * | 16 (100%) |
| Huber classification of right eye if affected – Type I * | 05 (100%) |
| *Does the affected eye amblyopic?* | |
| • Yes | 07 (35.0%) |
| • No | 13 (65.0%) |
| *Abnormal head position* | |
| • Yes | 05 (25.0%) |
| • No | 15 (75.0%) |
| *Clinical characteristics of the disease* | |
| • Esotropia | 06 (30.0%) |
| • Orthotropic | 14 (70.0%) |
| *Severe globe retraction* | |
| • Yes | 01 (05.0%) |
| • No | 19 (95.0%) |
| *Strabismus in primary position* | |
| • Yes | 06 (30.0%) |
| • No | 14 (70.0%) |
| *Is there associated abnormal eye movement?* | |
| • Yes | 04 (20.0%) |
| • No | 16 (80.0%) |
| *If yes, what is it?* | |
| • Upshoot | 03 (75.0%) |
| • downshoot | 01 (25.0%) |
| *History of previous ocular surgery* | |
| • Yes | 03 (15.0%) |
| • No | 17 (85.0%) |
| If yes, please specify – Medial rectus muscle recession OS | 03 (100%) |
| *Systemic congenital malformation* | |
| • Yes | 01 (05.0%) |
| • No | 19 (95.0%) |
| If yes, please specify – Mental retardation | 01 (100%) |
| *Doctor action plan* | |
| • Glasses | 11 (55.0%) |
| • Frequent follow up | 09 (45.0%) |
| • Observation | 08 (40.0%) |
| • Patching | 07 (35.0%) |
| • Need surgical intervention | 02 (10.0%) |

* 1 patient with both (right and left) affected eye has been distributed equally in Huber classification of eye.

patients had amblyopia in the affected eye due to anisometropia or hyperopic refractive error. 25% of all patients had an abnormal head position. 70% of the patients were orthotropic while 30% had Esotropia in the primary gaze postion. 70% of patients had a positive history of parents' consanguinity while 30% were negative. 30% of the patients had a positive family history of strabismus, either exotropia or exotropia. Severe globe retraction was found in one patient (05.0%). Four patients (20.0%) had associated abnormal eye movement. Out of these, three patients (75.0%) had upshoot while one patient (25.0%) had downshoot. Associated abnormal eye movement more common when the right eye is affected (p-0.035). Three patients (15.0%) had history of previous ocular surgery and had medial rectus muscle recession of the affected eye. None of the patients had other ocular congenital abnormality, systemic or congenital abnormality, except one who had learning disability. 75% of the patients had the disease since birth, 15% of them developed the disease from 1 month to 6 months and ten percent of them developed the disease about more than 6 months to one year (Table 1). Sixteen (80.0%) of our patients were from Al-Medina city while four (20.0%) of them were from different area of Al-Medina region (Al Hejeria, Al Henakia, Omluj, Yatmah).

In regard to the physician action plan, all patients were in observation with or without glasses and/or patching for amblyopia, except for two patients who were recommended surgical intervention due to abnormal head position. Three patients had previous history of medial rectus muscle recession in the affected eye. All were clinically clinically improved after surgery without any complications. Statistical analysis was done to see association between demographic and clinical characteristics versus affected eyes (right or left) shown in Table 3. Analysis revealed that among all important variables, only associated abnormal eye movement (p-0.035) found to be statistically significant. Associated abnormal eye movement (upshoot and downshoot) more common when the right eye is affected. Other variables included in the table shows no significant relationship in the two groups of the affected eye.

**Table 3**. Relationship between Socio-Demographic and Clinical Characteristics versus patients affected eye [(n=19)].

| Characteristics | Affected Eye[¥] | | P-value[∞] |
|---|---|---|---|
| | Left Eye [(n=15)] | Right Eye [(n=4)] | |
| Age in years | 07.8 ± 08.4 | 08.0 ± 12.7 | 0.357 |
| *Gender* | | | |
| • Male | 07 (77.7%) | 02 (22.2%) | 0.906 |
| • Female | 08 (80.0%) | 02 (20.0%) | |
| *Residency* | | | |
| • Al Madinah | 12 (80.0%) | 03 (20.0%) | 0.827 |
| • Outside Al Madinah | 03 (75.0%) | 01 (25.0%) | |
| *Parents Consanguinity* | | | |
| • Positive | 09 (69.2%) | 04 (30.8%) | 0.126 |
| • Negative | 06 (100%) | 0 | |
| *Positive Family History of Strabismus* | | | |
| • Yes | 05 (100%) | 0 | 0.179 |
| • No | 10 (71.4%) | 04 (28.6%) | |
| *Onset of the disease* | | | |
| • Since birth | 10 (71.4%) | 04 (28.6%) | 0.405 |
| • 1 month–6 months | 03 (100%) | 0 | |
| • >6 months–1 year | 02 (100%) | 0 | |
| *Does the affected eye amblyopic?* | | | |
| • Yes | 05 (71.4%) | 02 (28.6%) | 0.539 |
| • No | 10 (83.3%) | 02 (16.7%) | |
| *Abnormal head position* | | | |
| • Yes | 04 (80.0%) | 01 (20.0%) | 0.946 |
| • No | 11 (78.6%) | 03 (21.4%) | |
| *Strabismus in primary position* | | | |
| • Yes | 04 (80.0%) | 01 (20.0%) | 0.946 |
| • No | 11 (78.6%) | 03 (21.4%) | |
| *Is there associated abnormal eye movement?* | | | |
| • Yes | 01 (33.3%) | 02 (66.7%) | **0.035**[**] |
| • No | 14 (87.5%) | 02 (12.5%) | |

*Results are expressed as mean ± standard deviation, number and percentage (%).
[¥] 1 patient have both affected eyes since it has been excluded from the analysis of this table.
[∞] P-value has been calculated using Chi square test and Mann Whitney U test.
[**] Significant value.

## Discussion

In this study, we aimed to assess the clinical characteristics of DRS in Al-Medina region. The clinical characteristics of DRS in this study are consistent with the findings of similar studies done before. The present study found that males and females were equally affected 1:1 (50% for each) whereas the gender incidence in Khan and Oystreck study was (55%) females and (45%) males.[3] This may be due to our small sample size. The left eye was affected in 15 patients (75%). While 20% (4 patients) had their right eye affected, both genders were equal (two for each). Most of the studies are consistent with our findings, as (74–76%) were left eye affected and (20–25%) were right eye affected.[3,9,10,11] Bilateral DRS was only found in single male case (5%). Emilie Zanin et al. found that out of 94 patients, 16 (17%) of them were bilaterally affected, also Ayse Gul Kocak et al. reported that out of 21 patients 1 case was bilaterally affected as well which is consistent with our study.[10,11] Amblyopia present in 35% (7 patients) of our patients which is higher to other compered studies.[3,9,11] However, regarding the findings of eye deviation are different from other studies. Here we noted esotropia in primary gaze in 30% and while the rest of 70% were orthotropic with no exotropia present. Kanwar Mohan et al. found that exotropia was significantly more common than esotropia in unilateral DRS, and patients with unilateral type I DRS had no significant difference in the incidence of esotropia and exotropia.[9] This difference may be due to geographical variation or due to small sample size. The prevalence of having upshoot and severe globe retraction was comparable to other studies. Ayse Gul Kocak et al. found similar results that out of 21 patients, 4 patients have upshoot and one patient has globe retraction.[11] Regarding our finding that associated abnormal eye movement (upshoot and dwonshoot) more common when the right eye is affected, no other slimier studies statistically demonstrated the association between them before.

Parents' consanguinity was found positive in (70%) of the study sample. This interesting finding gives the importance of further evaluation of the association between consanguinity and the occurrence of DRS in our region supporting the result reported by Khan & Al-Mesfer.[12]

The main limitations of this study were the sample size: There was no coding mechanism to allow us to identify and collect cases of DRS. Hence, we collected cases prospectively by identifying the cases in the strabismus clinic during the six months period. This may explain the small sample size.

## Conclusion

This cross-sectional observational study is the first one that evaluates DRS in Al-Medina region. The prevalence of amblyopia in this study is higher than other compered studies. We found that parents' consanguinity may play a role in the occurrence of DRS as it is not considered as a rare disease in our region. Abnormal eye movement more common when the right eye is affected. Also, further studies are needed to establish the relationship between abnormal eye movement and the involvement of the right eye.

## Declaration of Competing Interest

The authors declared that there is no conflict of interest.

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