

The Incidence Of Cephalofacial Birth Defects

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The orthodontist is concerned with the health and appearance of the head and face. It has been found that orthodontic treatment can influence the configuration of the oral, dental and facial structures. The greatest benefits of treatment seem to be to the alignment and positioning of the teeth. The timing of treatment to cooperate with or to influence the direction of growth gives the greatest opportunity or promise of change of the other structures of the head and face. These changes are evident and have been recorded as occurring in both the hard and soft tissues. The orthodontist is sometimes faced with problems that he cannot correct completely. Some of these problems are the result of birth defects; others are acquired postnatally. A better understanding of the magnitude and complexity of these birth defects should be helpful in orthodontic treatment planning. It is felt that this knowledge will be as useful as is the knowledge of the frequency of the incidence of malocclusion. We will try to better delineate the margin between the deformities that are and are not amenable to orthodontic treatment.

MATERIALS AND METHODS

The scientific literature on the subject of birth defects and congenital malformations was reviewed. This review of the literature would adequately and accurately report the status of birth defects for the general population, es-

pecially for the North American Caucasian population. The review of the literature found a dearth of information on the incidence of birth defects among North American Negro population. No large or representative studies had been done. To supplement the information on the general population a survey was made of the birth records at two hospitals in the District of Columbia, namely, the D.C. General Hospital and Freedmen's Hospital. These hospitals were selected because they give a cross section of the Negro community in Washington, D.C. A large percentage of the children of the Negro upper and middle socioeconomic groups are born in Freedmen's Hospital; the great majority of the indigent and lower socioeconomic group are born at D.C. General Hospital. Negro children in the District of Columbia represent approximately 71% of the total live births and approximately 60% of these children were born at the two hospitals studied. It is felt that this group because of its homogeneous nature is representative of the North American Negro community and therefore that these data will be significant.

The birth records for a ten year period were examined at the two hospitals described above. The ten year periods selected were from September 1, 1952 to August 31, 1962 inclusive for Freedmen's and from January 1, 1952 to December 31, 1961 inclusive for the D.C. General Hospital. It was desired to have duplicate or similar periods for both hospitals but this was not possible because of record storage, filing, and other differing administrative policies at the two institutions. It was felt that the

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data would not be affected by the slight differences in the dates. The total number of records examined for the ten year periods are as follows:

Freedmen's Hospital	26,131
D.C. General Hospital	53,711
Total	79,842

The data recorded from each birth record examined were as follows:

Sex

Live or Stillbirth

Normal or Abnormal Birth

The name or description of the defect or malformation was copied from the medical chart. No attempt was made at interpretation at that time. After all records had been checked, consultations were held with an obstetrician and pediatrician on the staff of each of the two hospitals. These conferences were very valuable to an understanding of the terminology.

PRESENTATION OF DATA

The data will be presented in the following manner: first, data on the incidence of cephalofacial birth defects in the general population; secondly, data

on the incidence of cephalofacial birth defects obtained from the study of the birth records of North American Negro children in the District of Columbia; thirdly, comparisons will be made between the data.

The frequency of the incidence of birth defects has been reported by a number of authors. Table I lists some of these. The periods studied varied from one year to twelve years and the number of births studied varied from several thousand to over a million. In reviewing the literature and in conducting the survey of the birth records of Negro children this author found the terminology used in referring to this subject confusing, different authors used different terms, e.g., some authors refer to birth defects, others to congenital anomalies, still others to congenital malformations. This author will refer to these conditions as birth defects except where another author is quoted.

Table I gives a review of some of the studies conducted on the frequency of the incidence of birth defects during the period from 1945 to the present, 1963. This table is taken from the study of Mellin.

These studies were made either of

TABLE I
Incidence of Congenital Malformations
Summary of Literature

References	Year	Cases	Congenital Malformations	% of Total
DePorte & Parkhurst	1945	273,604	3,180	0.86
Hirst	1945	24,100	198	1.2
Murphy	1947	7,478	222	1.3
Lucy	1949	10,751	210	2.0
Miller	1950	4,095	66	1.6
Stevenson, S., et al	1950	29,024	677	2.3
Hendricks	1955	210,727	1,560	1.3
Shapiro, et al	1958	30,398	604	5.0
Gentry, et al	1959	1,242,744	16,369	0.75
Sesgin & Stark	1960	27,087	511	1.88
Mellin	1963	9,783	293	3.0

TABLE II
Incidence of Birth Defects
North American Negro Children

Births	Freedmen's Hospital	D. C. General Hospital	Totals
Live Births	26,181	53,711	79,842
Normal Births	25,502	49,685	75,187
Abnormal Births	629	4,026	4,655
Percentage of Total	2.46	7.49	5.83

the birth records, medical charts, or of birth certificates on file in the bureaus of vital statistics in various states and cities of the U.S.A. The number of births surveyed varied from 4095 to 1,242,744. The periods covered by these surveys vary also from one year to twelve years. The racial identities of the subjects used in these studies were mixed but most of the studies were of Caucasian children. The number of congenital malformations varied from a low of 66 in a study of 4095 births to a high of 16,369 in a study of 1,242,744 births. The percentages of malformations varied from a low of 0.75% to a high of 5.0%. These data represent the total incidence of all types of congenital malformation. It is felt that the range of variability of these frequencies is related to the different samples that constituted the various studies and to the differing classifications as to what constitutes a malformation or birth defect.

Table II presents the totals of a study conducted at two hospitals in the District of Columbia. These data are on the frequency of birth defects among North American Negro children. The birth records, i.e., medical charts, were studied and the diagnosis or description of the birth was copied verbatim with no attempt at interpretation at the time of examination. The totals show that 4655 or 5.83% of the 79,842 live births were abnormal, i.e., they

had one or more defects. Attention is called to the great differences in the total frequency of birth defects at the two hospitals. The D.C. General Hospital whose patients are of a lower socioeconomic group than those at Freedmen's Hospital have a higher incidence. Some answers that are advanced to explain these differences are the lack of adequate prenatal care, poor hygiene, malnutrition and a higher incidence of infectious diseases among the mothers of this group. The author plans to explore these factors in greater detail in a future study.

The above data are on the total incidence of birth defects. To give a somewhat more detailed view of the frequency of the incidence of individual defects Table III presents the ten most frequent congenital malformations. We refer here to congenital malformations rather than birth defects; for this grouping we have selected those children that were anatomically malformed. There seems to be less confusion and controversy in this classification of birth defects than in the broader classification which also includes birth injuries and diseases. The magnitude and nature of birth injuries and diseases seem to vary from one study to another, and many of these conditions either clear up spontaneously or respond rapidly to postnatal treatment.

This table utilizes the data of Stevenson, Worcester and Rice of the Harvard

TABLE III
Ten Most Frequent Congenital Malformations

CAUCASIAN CHILDREN		NEGRO CHILDREN	
Defect	Incidence	Defect	Incidence
Hypospadias	1:1001	Polydactyly	1:117
Polydactyly	1:1319	Hernia	1:199
Hydrocephaly	1:1451	Clubfoot	1:346
Mongolism	1:1935	Pre-Auricular Appendages	1:1331
Cleft Palate & Harelip	1:1935	Hydrocele	1:1331
Syndactyly	1:2073	Hemangioma	1:1377
Cleft Palate	1:2639	Nevi	1:1535
Congenital Heart Disease	1:4535	Tumors, Tags, Growths	1:1996
Anencephaly	1:6047	Hypospadias	1:2101
Clubfoot	1:9675	Supernumerary Nipples	1:2851

School of Public Health on the incidence of congenitally malformed births at the Boston Lying-In Hospital during the years 1930 through 1941. These data were chosen as a representative study of the North American Caucasian population. In this study 29, 024 records were studied, 677 or 2.3% had congenital malformations. The racial identity of this group was Caucasian except for 19 children, 18 Negroes and 1 Chinese. It was felt that this small mixture would not significantly affect these data. The data of the District of Columbia study were chosen as representative of the North American Negro population. The ten most frequently occurring congenital malformations were selected from both groups. These are listed in the order of decreasing frequency. The table shows the differences that were found most frequently in these two groups. The most common malformation in the Caucasian group was hypospadias, whereas the most common malformation of the Negro group was polydactyly. It will be noted that five of the most frequent malformations of the Caucasian group, namely, mongolism, cleft palate alone, cleft palate and harelip, hydrocephaly, and anencephaly are defects of the head and face, whereas only one of the most fre-

quent malformations of the Negro group, namely, preauricular appendages is a defect of the head and face. With these tables and comments we conclude this brief glimpse of the total incidences of birth defects.

BIRTH DEFECTS OF THE HEAD AND FACE

The main purpose of this study was to gather data that would be useful to the orthodontist, therefore, we shall concentrate henceforth on the birth defects of the head and face. Some of these defects are amenable to orthodontic treatment and some are not. We shall try to discuss in greater detail those defects which require orthodontic treatment. Some will require the services of the general and oral surgeon, the prosthodontist, the speech therapist, the pediatrician and other medical and dental specialties. This "team" approach to the treatment of these children has been very successful.

The frequency of the incidence of single defects of the head and face of a North American Caucasian group as reported by Stevenson, et al is shown in Table IV. Eight defects are listed in this table — of this group four or one-half of these defects have either oral, facial or dental problems associated with them. This group, namely, mongolism, cleft

TABLE IV
Incidence of Single Defects
of the Head & Face
North American Caucasian Children

Defect	Incidence
Anencephaly	1:605
Hydrocephaly	1:1456
Mongolism	1:1935
Cleft Palate & Harelip	1:1935
Cleft Palate	1:2639
Meningocele	1:2902
Harelip	1:3628
Microcephaly	1:14,512

palate and harelip, cleft palate alone, and harelip alone, occurred in this group of Boston children once in approximately every 2000 to 3500 births. The other four defects listed, namely, anencephaly, hydrocephaly, meningocele, and microcephaly may or may not have oral, facial or dental defects associated with them. This latter group of defects is not usually seen by the orthodontist for treatment, in fact, many of

TABLE V
Incidence of Single Defects
of the Head & Face
North American Negro Children

Defect	Incidence
Cephalohematoma	1:570
Pre-Auricular Appendages	1:1331
Craniotabes	1:2158
Mongolism	1:3071
Dentia Praecox	1:3471
Cleft Palate	1:4697
Cleft Lip & Palate	1:6654
Ankyloglossia	1:6654
Hydrocephaly	1:7258
Microcephaly	1:7258
Macrocephaly	1:11,406
Dolichocephaly	1:11,406
Cleft Lip	1:11,406
Maxillary Gingival Clefts	1:13,307
Nasal Obstruction	1:15,968
High Arched Palate	1:19,961
Macroglossia	1:19,961

these children die at a very early age. The children in the first group, with the rare exception of mongolism, are frequently seen by the orthodontist.

The incidence of single defects of the head and face of the North American Negro child is shown in Table V. These data are taken from the District of Columbia study. Seventeen defects are listed with frequencies of incidence of from 1:570 for cephalohematoma to 1:19,961 for macroglossia. Nine of this group of defects have oral, facial or dental problems associated with them. These defects are, namely, mongolism, dentia praecox, cleft lip and palate, cleft palate alone, ankyloglossia, cleft lip alone, maxillary gingival clefts, high arched palates, and macroglossia. These defects frequently require some orthodontic treatment. Here again, as with the previous group of children, the orthodontic treatment is often only one phase of the necessary medical and dental care required by these children. The remaining eight of the defects listed in Table V may or may not have accompanying oral, facial or dental problems associated with them.

We list in Table VI the multiple defects of the head and face found in the North American Negro group. These defects occurred very rarely, each group listed occurring only once. This table is included because many of these conditions can require orthodontic treatment as a part of their overall therapy. No similar table is presented for North American Caucasian children — the frequency was too rare.

SUMMARY

The literature on the subject of the frequency of the incidence of birth defects for the past twenty years was reviewed. A birth defect was found to occur in a range of from one to five percent of all live births. The data in these studies were taken either from

TABLE VI
Multiple Defects of the Head and Face
North American Negro Children

Micrognathia	Hydrocephaly
High Arched Palate	Meningitis
Dentia Praecox	Hydrocephaly
Cyst Upper Left Gingiva	Hematoma
Dentia Praecox	Meningocele
Tumor on Lower Jaw	Extra Cranial Mass
Cleft Palate	Hydrocephaly
Congenital Cataracts	Meningitis
Cleft Lip & Palate	Brain Abscess due to
Micro-Opthalmia	Pseudomonas Aeruginosa
Mongolism	Skin Tag on Ear Anterior
Soft Palate Defect	to Pinna
Upper Gum Cleft	Left Facial Paralysis
Mongolism	Cephalohematoma
Dentia Praecox	Gingival Cysts
Hydrocephaly	Milia Over Nose
Osteogenesis Imperfecta	Cysts on Gum
of the Head	

birth records, medical charts, or from birth certificates on file in the bureaus of vital statistics in various cities and states of the U.S.A. These data were taken from mixed racial groups that were predominantly of Caucasian children. It was found that there was a dearth of information in the literature on the frequency of the incidence of birth defects among North American Negro children. To supplement this information a survey was conducted of the birth records for a ten year period at two hospitals in the District of Columbia. The frequency of the incidence of birth defects among these Negro children was found to be approximately six percent.

The ten congenital malformations occurring most frequently were reported. The defects occurring in a group of Boston children that were predominantly Caucasian were compared with the defects occurring in the District of Columbia group of Negro children. There were differences in the malformations occurring most frequently;

only three of the defects, polydactyly, clubfoot, and hypospadias, were found to occur in both groups, and the frequencies of these defects varied considerably.

After a brief presentation of the above information on the total incidence of birth defects we concentrated on defects of the head and face. We attempted to present information that would be useful to the orthodontist by discussing in greater detail those defects that would require orthodontic treatment as a portion of the overall therapy necessary to either correct or improve these conditions. It was found that approximately one half of the birth defects of the head and face in both the Caucasian and Negro groups had either oral, facial or dental problems associated with them. As some of these defects occur as frequently as once in every few hundred to several thousand live births, they have a definite importance to the dentist and orthodontist.

CONCLUSIONS

1. A large number of children have

birth defects, but there is considerable variation in the frequency of the incidence of these defects. It is felt that some of this variation may be due to several factors, e.g., different methods of studying and reporting these defects, a lack of a uniform terminology and classification.

2. There are different frequencies in the incidence of these defects for different geographic locations.

3. The racial identity of the sample studied seems to have an influence on the frequency of the incidence of the various birth defects. The defects occurring most frequently are different for North American Caucasian and Negro children. In the ten malformations occurring most frequently only three appeared in both groups and the frequencies of these varied considerably.

4. There is variability in the frequency of the incidence of birth defects of the head and face as well as other parts of the body.

5. There are birth defects of the head and face that require orthodontic treatment for their correction or improvement. There are also birth defects that are not amenable to orthodontic treatment. The orthodontist either alone or as a member of the "team" that is needed to treat many of these children can make a real contribution toward helping them enjoy a healthier and happier life.

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REFERENCES

1. Davis, J. S.: The Incidence of Congenital Clefts of the Lip and Palate. *Ann. Surg.*, 80: 363, 1934.
2. DePorte, J. V., and Parkhurst, E.: Congenital Malformations and Birth Injuries Among Children Born in New York State, Outside of New York City, in 1940-1942, *New York State J. Med.*, 45: 1097, 1950.
3. Mellin, G. W.: *Birth Defects*, Philadelphia, Lippincott Chap. 1, pp. 1-14, 1961.
4. Gentry, J. T., Parkhurst, E., and Bulin, G. V., Jr.: An Epidemiological Study of Congenital Malformations in New York State, *Am. J. Pub. Health*, 49: 497, 1950.
5. Grace, L. G.: Frequency of Occurrence of Cleft Palate and Harelip, *J. Dent. Res.*, 22: 495, 1943.
6. Harris, L. E., and Steinberg, A. G.: Abnormalities Observed During the First Six Days of Life in 8,716 Live Born Infants, *Pediatrics*, 14:314, 1954.
7. Hendricks, C. H.: Congenital Malformations: Analysis of the 1953 Ohio Records, *Obstet. Gynec.*, 6: 1952, 1955.
8. Hirst, J. C.: *Cyclopedia of Medicine, Surgery, and Specialties*, Vol. 10, p. 189, Philadelphia, F. A. Davis, 1945.
9. Hixon, E. H.: A Study of the Incidence of Cleft Lip and Cleft Palate in Ontario, Canada, *J. Pub. Health*, 42:508, 1951.
10. Ivy, R. H.: The Influence of Race on the Incidence of Certain Congenital Anomalies, Notably Cleft Lip — Cleft Palate, *Plast. & Reconstruct. Surg.*, 30: 581-585, Nov., 1962.
11. Lucy, R. E.: A Study of Congenital Malformations, *J. Lancet*, 69: 80, 1949.
12. McIntosh, R., Merritt, K. K., Richards, M. B., Samuels, M. H., and Bellows, M. T.: The Incidence of Congenital Malformations: A Study of 5,946 Pregnancies, *Pediatrics* 14: 505, 1954.
13. Miller, H. C.: Scope and Incidence of Congenital Abnormalities, *Pediatrics*, 5: 320, 1950.
14. Sesgin, M. Ziya, and Stark, R. B.: The Incidence of Congenital Defects, *Plast. & Reconstruct. Surg.*, 27: 261-266, 1959.
15. Shapiro, R. H., Eddy, W., Fitzgibbon, J., and O'Brien, G.: Incidence of Congenital Anomalies Discovered in the Neonatal Period, *Am. J. Surg.*, 96: 396, 1958.
16. Slaughter, W. B., and Phair, G. M.: A Complete Cleft Palate Program, *J. Speech and Hearing Disorders*, 17: 123, 1952.
17. Stevenson, S. S., Worcester, J. and Rice, R. G.: 677 Congenitally Malformed Infants and Associated Gestational Characteristics, Part I: General Considerations, *Pediatrics*, 6: 37, 1950. Part II: Prenatal Factors, *Pediatrics*, 6:208, 1950.
18. Wallace, H. M., Baumgartner, L., and Rich, H.: Congenital Malformations and Birth Injuries in New York City, *Pediatrics*, 12: 525, 1953.
19. Wallace, H. M., Hoenig, L. and Rich, H.: Newborn Infants with Congenital Malformations or Birth Injuries, *Am. J. Dis. Child.*, 91: 529, 1956.
20. Warkany, J.: Etiology of Congenital Malformations, *Advances Pediat.*, 2: 1, 1947.